IN THE UNITED STATES PATENT AND TRADEMARK OFFICE

Application of:	Docket No.30307A
KNOLL, JOAN et al.	
Serial No. :09/854,867	Group Art Unit No. 1634
Filed: May 14, 2001	
SINGLE COPY GENOMIC HYBRIDIZATION PROBES AND METHOD OF GENERATING SAME	Examiner: C. Myers

Commissioner of Patents
Alexandria, VA 22313-1450

Sir:

PETITION TO THE COMMISSIONER
UNDER 37 CFR § 1.84(a)(2) TO ACCEPT PHOTOGRAPHS

Applicant hereby petitions the Commissioner of Patents and Trademarks, under 37 CFR § 1.84(a)(2) to accept these photographs into the above-referenced application. Enclosed herewith are three (3) sets of photographs to be entered into the above-referenced application; along with one set of black and white copies that accurately depicts, to the extent possible, the subject matter shown in the color drawings. A check in the amount of $130.00 in accordance with § 1.17 (h). Applicant requests favorable consideration of this petition

Any additional fees which are due in connection with this Petition should be applied against our Deposit Account No. 19-0522.

Respectfully submitted,

By _____

Tracey S. Truitt, Reg. No. 43,205
HOVEY WILLIAMS LLP
2405 Grand Boulevard, Suite 400
Kansas City, Missouri 64108
816/474-9050

ATTORNEYS FOR APPLICANT(S)

Fig. 1

Fig. 2

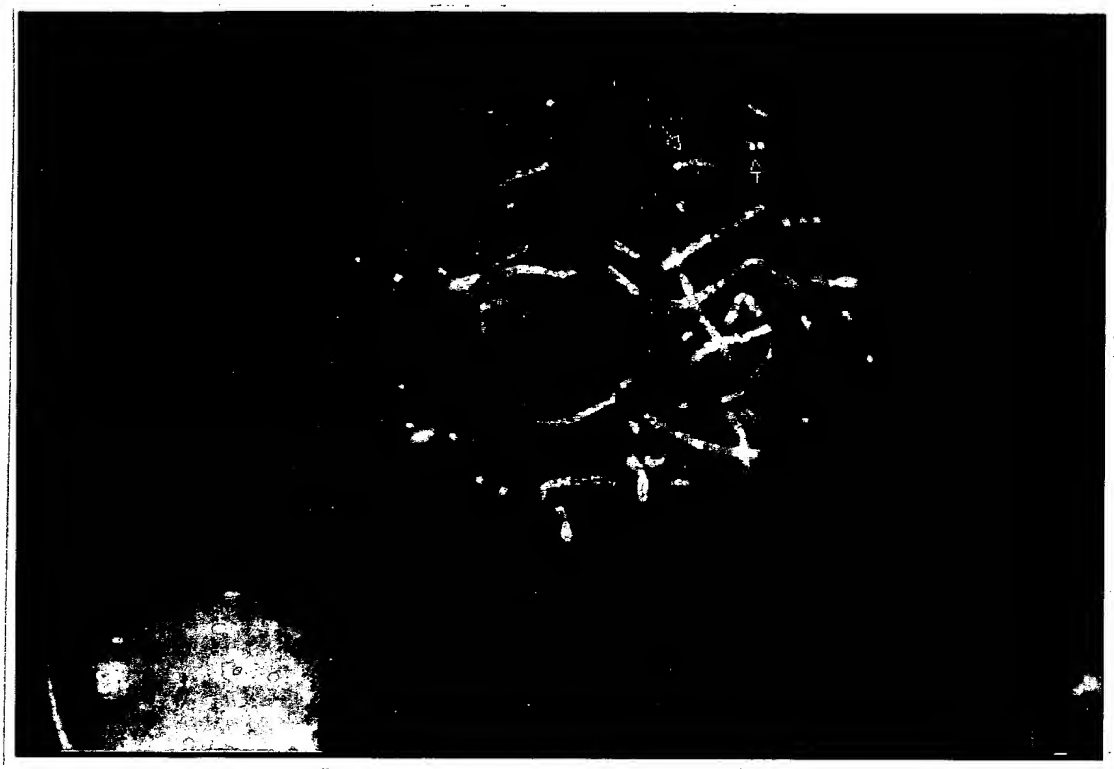

Fig. 3

Fig. 4

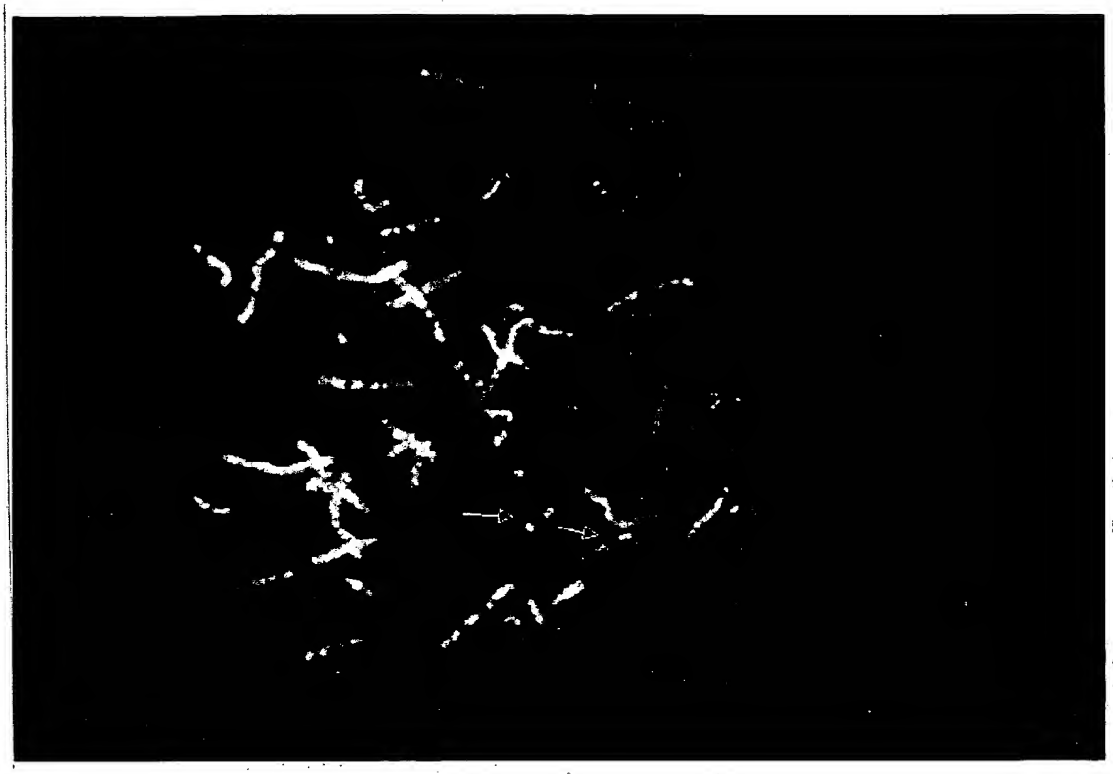

Fig. 5

Fig. 6

Fig. 7

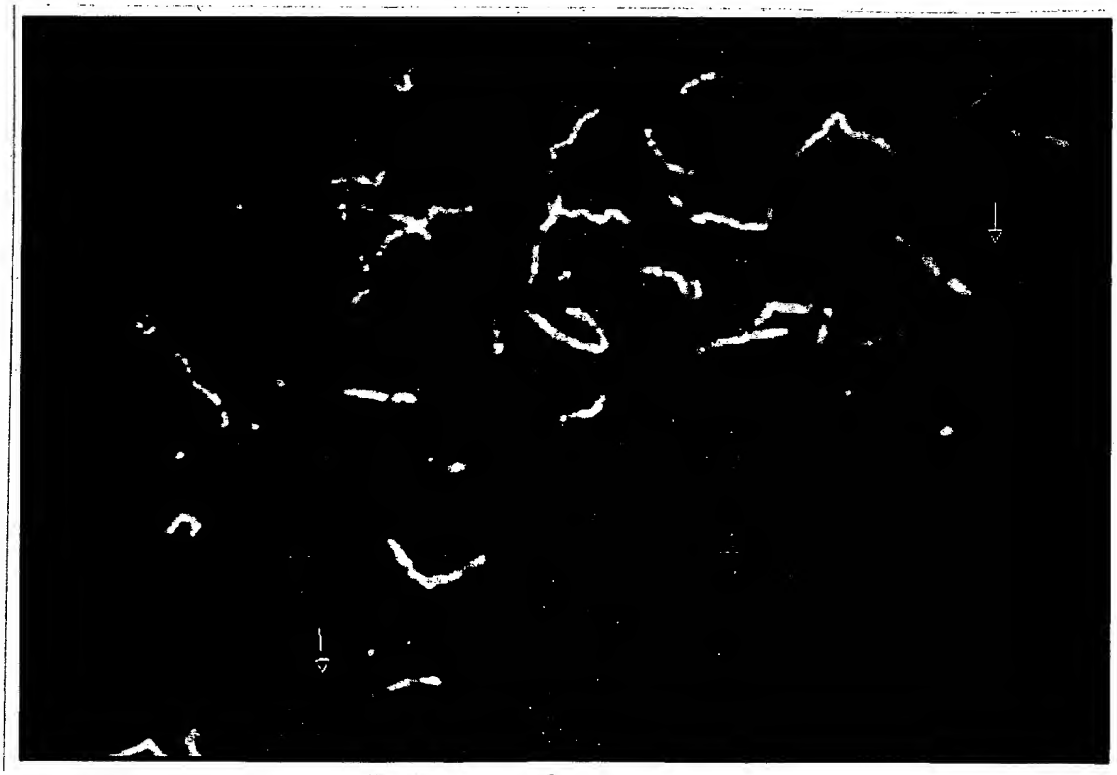

Fig. 8

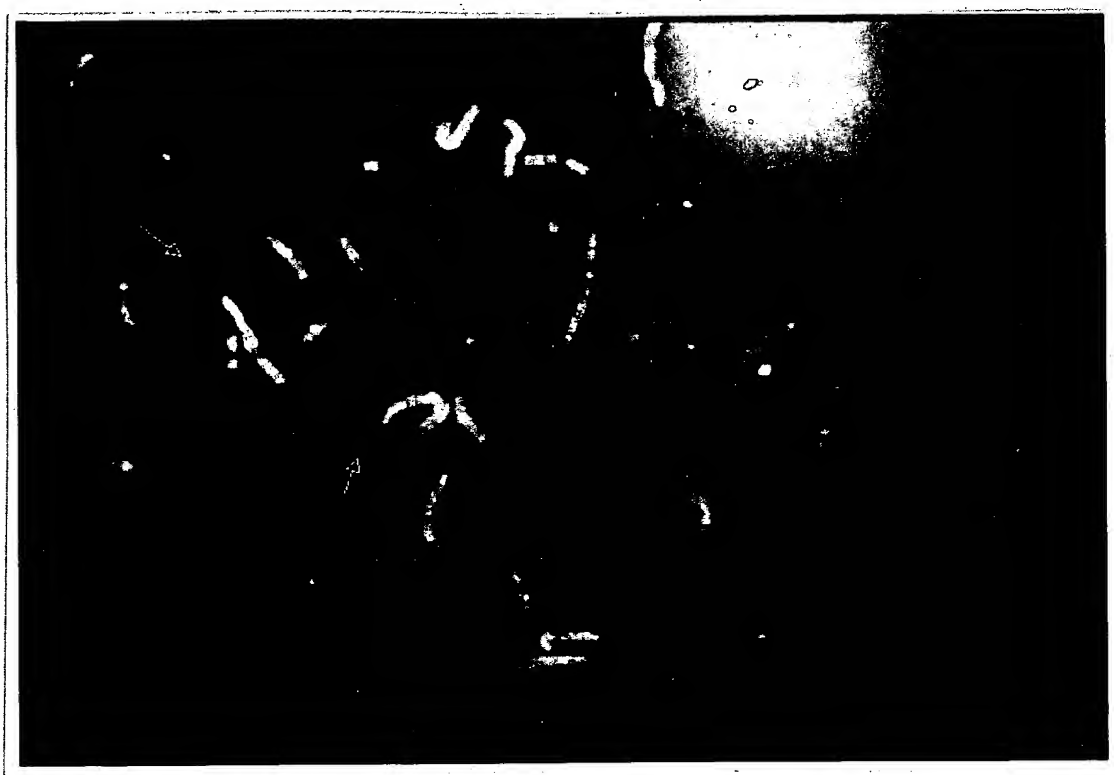

Fig. 9

Fig. 10

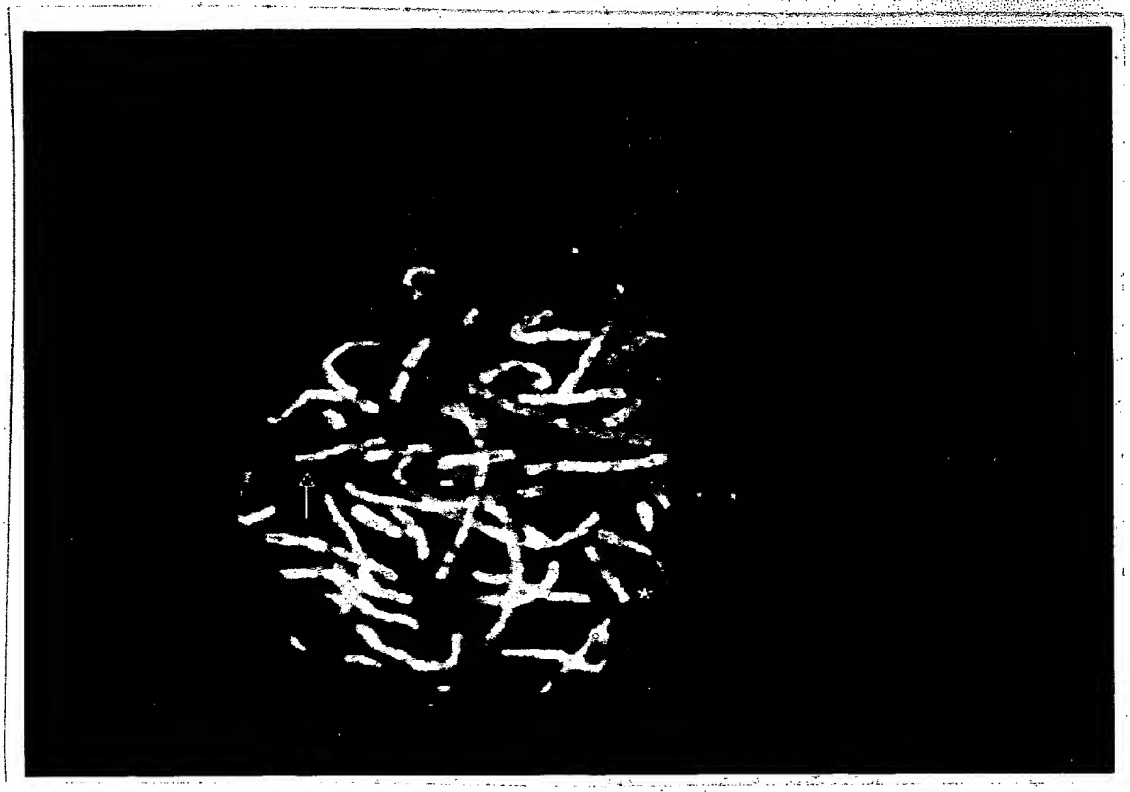

Fig. 11

Fig. 12

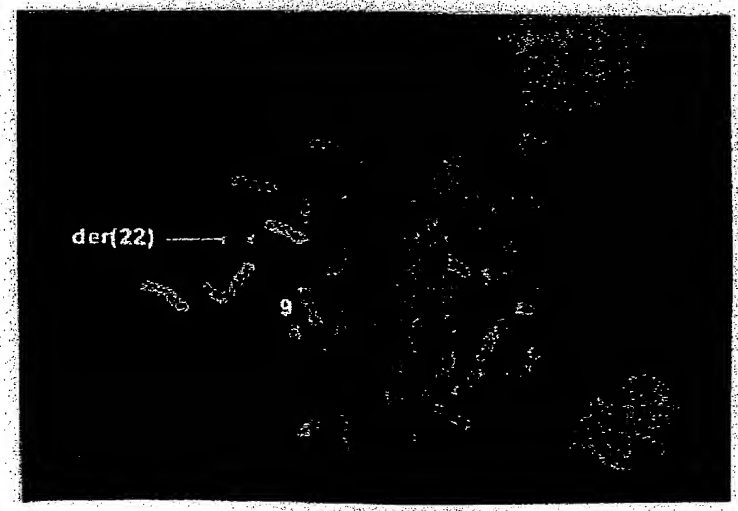

Fig. 15

Fig. 16

Figs 15 16.jpg

PETER K. ROGAN, Ph.D.
Laboratory of Human Molecular Genetics
Department of Medical Research
Children's Mercy Hospital and Clinics
2401 Gillham Rd.
Kansas City, MO 64108
Tel: (816) 960-4820
Fax: (816) 753-1307
E-mail: progan@cmh.edu
WWW: http://www.sce.umkc.edu/~roganp/

EDUCATION

1979	B.A.	Biophysics	Johns Hopkins University
1980	D.E.A.	Cellular Biology	Université de Grenoble, France
1983	M.Phil.	Molecular Biophysics and Biochemistry	Yale University
1987	Ph.D.	Molecular Biophysics and Biochemistry (Human Genetics)	Yale University (Advisor: S. M. Weissman, M.D.)

WORK EXPERIENCE AND POSTGRADUATE TRAINING

1979 - 1980	Research Assistant, Biophysics Laboratory	Institut Laue-Langevin Grenoble, France
1984 - 1986	Consultant, Computational Molecular Biology	Dept. Human Genetics, Yale University School of Medicine
1987 - 1990	Postdoctoral Fellow, Laboratory of Eukaryotic Gene Expression	ABL/Basic Research Program National Cancer Institute, NIH
1990 - 1996	Assistant Professor, Dept. Pediatrics, Div. Genetics	College of Medicine, The Pennsylvania State University
1993	Participant, Genetic Linkage Analysis Course (Director, J. Ott)	College of Physicians and Surgeons, Columbia University
1995 - present	President	Phylogenetix Laboratories, Inc.
1996 - 1999	Associate Professor, Human Genetics	MCP Hahnemann School of Medicine
1999 - 2004	Associate Professor, Pediatrics	School of Medicine, University of Missouri-Kansas City
2001 - 2004	Associate Professor, Computer Science	School of Interdisciplinary Computing and Engineering, University of Missouri-Kansas City
2002- present	Adjunct Associate Professor	Department of Chemistry, University of Kansas
2004 - present	Professor, Pediatrics and Computer Science	Schools of Medicine and Computer Science & Engineering, University of Missouri-Kansas City

HONORS, AWARDS AND PROFESSIONAL RECOGNITION

Bourse d'Etudes (Government of France), 1979-1980
National Research Service Predoctoral Award, 1980-1984
Postdoctoral Fellowship Award, ABL-Basic Research Program, 1987-1988

Scientific Conference Coordinator: Prader-Willi Syndrome Association (National), 1992
Basil O'Conner Starter Scholar, March of Dimes, 1992-1993
Shannon Award, National Institutes of Health, 1992-1993
Elected to Human Genome Organization, 1992
Director, March of Dimes-Central PA Molecular Diagnostic Laboratory, 1993-1996
March of Dimes Birth Defects Foundation Community Service Award, 1994
Honors, Pennsylvania Society of Professional Engineers -Harrisburg chapter, 1996
Who's Who in the East, 1997
Translational Research Award, Children's Cancer Group, 1999
Professorship in Pediatric Molecular Genetics, Children's Mercy Hospital, UMKC School of Medicine, 1999
External Advisory Board; Pharmacogenetics of Anticancer Agents (PharmGKB network, NIGMS PHS), 8/2003-.

PROFESSIONAL AFFILIATIONS
Sigma XI, 1987- ; American Association for the Advancement of Science, 1984- ; American Society of Human Genetics, 1990- ; American Association for Cancer Research, 1996- ; Human Genome Organization, 1992- ; Mutation Database Association 1997-; International Society for Computational Biology 2003-

GRANTS REVIEWED
Louis B. Leakey Foundation, US; National Science Foundation, US; Action Research Charity, Great Britain; National Heart Lung and Blood Institute, NIH; University of Kansas, Department of Pharmaceutical Chemistry Training Grant, PHS 9/1999; University of Missouri Research Board, 3/2003; Kansas City Area Life Sciences Institute 8/2003.

MANUSCRIPTS REVIEWED
Nucleic Acids Research; Genomics; The American Journal of Medical Genetics; American Journal of Human Genetics; Journal of Medical Genetics; Clinical Genetics; Cytogenetics and Cell Genetics; Journal of Clinical Endocrinology and Metabolism; Acta Pediatrica; Journal of the American Medical Association; Journal of Pediatric Hematology and Oncology; Human Genetics

INVENTIONS AND PATENTS
Invention Disclosure 91-1088 (PSU); Detection of Active Genomic Transcription Templates by Synthetic Methylation in Vivo; Disclosure Date: 10/18/91; Status: Disclosed 11/91

Invention Disclosure 92-1151 (PSU); Human Genetic Mapping with Recombinant Disomic Chromosomes; Disclosure Date 6/15/92; Status: Disclosed 11/13/92 - Presented to American Society of Human Genetics

Invention Disclosure 93- 1226 (PSU) [1]Method for rapid identification of prokaryotic and eukaryotic organisms; US Patent # 5,849,492.

Invention Disclosure 94- 1339 (PSU); A Method to Define the Chromosomal Location of Disease Genes that Cause Recessive Congenital Disorders; Disclosure Date: 4/18/94

Invention Disclosure 94- 1440 (PSU/NIH); Computational analysis of nucleic acid information defines binding sites; Status: US Patent # 5,867,402, licensed 3/98

---

[1]Rights to this invention have been acquired by Phylogenetix Laboratories, Inc.

Invention Disclosure 95- 1466 (PSU); 15qllql3 End Clones for Diagnostic Testing of Chromosome Rearrangements and Mutations; Disclosure Date: 4/26/95; Status: Disclosed 10/21/94

Invention Disclosure 95- 1551 (PSU); General Method of Detection of Minimal Residual Disease on Relapse after Non-Autologous Bone Marrow Transplant in Leukemia Patients; Disclosure Date: 12/15/95

Invention Disclosure 97- 0296 (Allegheny General Hospital); Information theory-based analysis of splice junction mutations in hereditary non-polyposis colon cancer; Disclosure Date: 8/1/97; Status: Disclosed 10/24/97

Invention Disclosure 00-0001 (Children's Mercy Hospital); Selection and generation of single-copy genomic probes for hybridization; Disclosure Date: 3/24/00; Status: Patent application Ser. No. 09/573,080, filed 5/16/00; Allowed 8/13/03; US Patent # 6,828,097 (12/7/04); Continuation application, filed 2/18/04.

Invention Disclosure 01-0001 (Children's Mercy Hospital); Single copy probes and method of generating same (Continuation-in-part); Disclosure Date: 4/7/00; Status: US Patent App. Ser. No. 09/854,867, filed 5/14/01; PCT/US01/15674.

Invention Disclosure 01-0002 (Children's Mercy Hospital); Subtelomeric DNA probes and method of producing same; Disclosure Date: 9-15-02; Status: US Patent App. Ser. No. 60/415,345, filed 9/30/02. Improved Sub-telomeric DNA Probes and Method of Producing Same, filed 07/2/03. Subtelomeric DNA probes and method of producing same, PCT\US03\31170, WO 2004/029283 A2, US Patent App. Ser # 10\676,248, filed 9/30/03.

Invention Disclosure 01-0004 (Children's Mercy Hospital); Computational selection of probes for localizing chromosome breakpoints in genetic diseases and cancer; Disclosure Date: 4/15/04; Status: US Patent App. Ser #60/557,007, filed 3/26/04.

APPROVED CLINICAL PROTOCOLS

Study Chair, B957: Genetic Etiology of Acute Leukemia in Children with Down Syndrome, Children's Oncology Group.

Study Committee, C297101: Therapy for Children with Down syndrome and acute leukemia, Children's Oncology Group.

PUBLICATIONS (in chronological order)

1. Wilson SR, Corvan PJ, Seiders RP, Hodgson DJ, Brookhart M, Hatfield WE, Miller JS, Reiss AH, Rogan PK, Gebert E, Epstein AJ : « The Structure and Magnetic and Electrical Conductivity Properties of the Charge Transfer Compound 1,1-Dimethylferrocenium Bis(tetracyanoquinodimethane), $(CH_3C_5H_3)_2Fe(TCNQ)_2$" in *Molecular Metals*, ed. W.H. Hatfield, Plenum Press, p 407-414ff, 1979.

2. Rogan PK, Williams GJB: The structure of the dihydrofolate reductase inhibitor 2,4,6-triamino-5-chloroquinazoline. Acta Cryst *B36*:2358-2362, 1980.

3. Rogan PK, Zaccai G: Hydration in purple membrane as a function of relative humidity. J Mol Biol *145*:281-284, 1981.

4. Mottez E, Rogan PK, Manuelidis LM: Conservation of the 5' region of the long interspersed mouse L1 repeat: Implications of comparative sequence analysis. Nucl Acids Res *14*:3119-3135, 1986.

5. Rogan PK: Restriction mapping by preferential ligation of adjacent digestion fragments. Nucl Acids Res *14*:9219, 1986.

6. Rogan PK: A study of a major long interspersed DNA repeat family common to rodents and primates. Ph.D. dissertation. Yale University, 1987.

7. Rogan PK, Pan J, Weissman SM: A complete L1 repeat element in the human ε-$^{G}\gamma$ globin gene intergenic region: Sequence analysis and concerted evolution within this family. Mol Biol Evol *4*:327-343, 1987.

8. Rogan PK, Salvo JJ: Molecular genetics of pre-Columbian South American Mummies. In: Molecular Evolution. UCLA Symposium on Molecular and Cellular Biology, *122*:223-234, 1990.

9. Rogan PK, Salvo JJ: Study of nucleic acids isolated from ancient remains. Yearbook Phys Anthropol *33*:195-214, 1990.

10. Rogan PK, Higgins DR: Screening UV-sensitive mutants with the Stratalinker® UV crosslinker. Strategies *4*:53-54, 1990.

11. Rogan PK: Research report: Ancient DNA studies. Soc. Arch. Sci. Bull. *14*:26-27, 1991.

12. Lemkin PF, Rogan PK: Automatic detection of noisy spots in two-dimensional Southern blots. Appl and Theor Electrophor *2*:141-149, 1991.

13. Rogan PK, Lemkin PL, Klar AJS, Singh J. Strathern JN: Two-dimensional agarose gel electrophoresis of restriction-digested genomic DNA. Methods: A Companion to Methods in Enzymology *3(2)*: 91-97, 1991.

14. Mascari MJ, Gottlieb W. Rogan PK, Butler MG, Waller DA, Armour JAL, Jeffreys AJ, Ladda RL, Nicholls RD: The frequency of uniparental disomy in Prader-Willi syndrome. New Eng J Med *326*:1599-1607, 1992.

15. Mascari MJ, Rogan P. Gannutz L, McCurdy M, Croft C, Lichty T. Ladda R: Pseudomosaicism Trisomy 15 in amniocytes: Concern about the possibility of uniparental disomy in the fetus. J Genet Counsel *1*:328-329, 1992.

16. Ladda RL, Rogan PK: Prader-Willi Syndrome Association Seventh Annual Scientific Meeting. Dysmorph Clin Genet *6(2)*: 64-65, 1992.

17. Ramer JC, Eggli K, Rogan PK, Ladda RL: Identical twins and Weissenbacher -Zweymuller Syndrome and neural tube defect. Am J Med Genet *45*:614-618, 1993.

18. Mowrey PN, Chorney M, Lerman M, Zbar B. Latif F. Rogan PK, Ladda RL: Clinical and molecular analysis of del 3p25-pter syndrome. Am J Med Genet *46*:623-629, 1993.

19. Ladda R. Zonana J. Ramer JC, Mascari MJ, Rogan PK: Congenital contractures, ectodermaldysplasia, cleft lip/palate and developmental impairment: A distinct syndrome. Am J Med Genet *47*:550-555, 1993.

20. Krizkova L, Sakthivel R. Olowe SA, Rogan P. Floros J: Human SP-A: Genotype and single strand conformation polymorphism analysis. Am J Physiol (Lung Cell Mol Physiol *10*): L519-L527, 1994.

21. Consevage MW, Salada GC, Baylen BG, Ladda RL, Rogan PK: A new missense mutation, Arg719Gln, in the β-cardiac heavy chain myosin gene in patients with familial hypertrophic cardiomyopathy. Hum Mol Genet *3*:1025-1026, 1994.

22. Woodage T. Prasad M, Dixon JW, Selby RE, Romain DR, Columbano-Green LM, Graham D, Rogan PK, Seip JR, Smith A, Trent RJ: Bloom syndrome and maternal uniparental disomy for chromosome 15. Am J Hum Genet *55*:74-80, 1994.

23. Gabriel J. Gottlieb W. Rogan PK, Saitoh S. Nicholls RD: A common insertion/ deletion polymorphism in the Prader-Willi syndrome minimal critical region. Hum Mol Genet *3*:1912, 1994.

24. Zaragoza MV, Jacobs PA, Rogan P. Sherman S. Hassold T: Non-disjunction of human acrocentric chromosomes. Human Genet *94*:411-417, 1994.

25. Rogan PK. Butler MG: Atypical clinical findings in PWS patients: A survey. Prader-Willi Perspect *2(4)*: 13-16, 1994.

26. Orr GA, Rogan PK: Development of a genetic probe database as a shared institutional resource. Computer Prog and Meth in Biomed *46*:35-39, 1995.

27. Rogan PK, Schneider TD. Using information content and base frequencies to distinguish mutations from genetic polymorphisms in splice junction recognition sites. Hum Mutation *6*:74-76, 1995.

28. Hess EJ*, Rogan PK*, Domoto M, Tinker DE, Ladda RL, Ramer JC: Absence of linkage of apparently single gene mediated ADHD with the human syntenic region of the mouse mutant coloboma. Am J Med Genet *60*:573-579, 1995.

29. Kauffman EJ, Gestl EE, Kim DJ, Walker C, Hite JM, Yan G. Rogan PK, Johnson SL, Cheng KC: Microsatellite-centromere mapping in the zebrafish *(Danio rerio)*. Genomics *30*:337-341, 1995.

30. Rogan PK, Close P. Blouin J-L, Seip JR, Gannutz L, Ladda RL, Antonarakis SE: Duplication and loss of chromosome 21 in two children with Down Syndrome and acute leukemia. Am J Med Genet *59*:174-181, 1995.

31. Rogan PK, Salvo JJ, Stephens RM, Schneider TD: Design of universal polymerase-chain reaction primers for amplification of 28S rDNA. In: Visualizing Biological Information, CA Pickover (ed). World Scientific, River Edge NJ, 1995.

32. Rogan PK, Driscoll DJ, Papenhausen PR, Johnson VP, Raskin S. Woodward AL, Butler MG. Distinct genotypes in Ring 15 and Russell-Silver syndromes. Am J Med Genet *61*: 10-15, 1996.

33. Veletza SV, Rogan PK, TenHave T. Floros J: Ethnic differences in allelic distribution at the human pulmonary surfactant protein B gene locus (SP-B): Relation to respiratory distress syndrome. Exp Lung Res *22*: 489-494, 1996.

* Co-principal authors

34. Consevage MW, Seip JR, Belchis DA, Davis AT, Baylen BG, Rogan PK: Novel association of a mosaic chromosomal 22qll deletion with hypoplastic left heart syndrome. Am J Cardiol 77: 1023-1025, 1996.

35. Rogan PK. Butler MG: Atypical clinical findings in PWS patients: Results of a survey. Prader-Willi Perspectives 4: 3-6, 1996.

36. Belchis DA, Meece CA, Benko FA, Rogan PK, Williams RA, Gocke CD: Loss of heterozygosity and microsatellite instability at the retinoblastoma locus in osteosarcomas. Diag Mol Pathol 5: 214-219, 1996.

37. Saitoh S, Buiting K, Rogan PK, Buxton JL, Driscoll DJ, Arnemann J, Fonig RK, Malcolm S, Horsthemke B, Nicholls RD: Minimal definition of the imprinting center and fixation of a chromosome 15qll-ql3 epigenotype by imprinting mutations. Proc Natl Acad Sci USA *93*:7811-7815, 1996.

38. White LM, Rogan PK, Nicholls RD, Wu Baylin, Korf B. Knoll JHM: Allele-specific replication of 15qll-ql3 loci: A diagnostic test for detection of uniparental disomy. Am J Hum Genet *59*:423-430, 1996.

39. Vgontzas AN, Kales A, Seip J. Mascari MJ, Bixler EO, Myers DC, VelaBueno A, Rogan PK: Relationship of sleep abnormalities to patient genotypes in Prader-Willi syndrome. Am J Med Genet (Neuropsychiatric Genetics) *67*: 478-482, 1996.

40. Floros J, DiAngelo S, Koptides M, Karinch AM, Rogan PK, Nielsen H, Spragg RG, Watterberg K, Deiter G: Human SP-A locus: Allele frequencies and linkage disequilibrium between the two surfactant protein A genes. Am. J. Respir. Cell and Mol. Biol. *15*: 489-498, 1996.

41. Nicholls RD, Jong MTC, Glenn CG, Gabriel J, Rogan PK, Driscoll DJ, and Saitoh S: Multiple imprinted genes associated with Prader-Willi syndrome and location of an imprinting control element. Acta Genet. Med. Gemellol. *45*: 87-89, 1996.

42. Mowery-Rushton PA, Hanchett JM, Zipf WB, Rogan PK, Surti U: Identification of mosaicism for paternally derived deletions in Prader-Willi syndrome using fluorescent *in situ* hybridization. Am. J. Med. Genet. *66*: 403-412, 1996.

43. Vgontzas AN, Bixler EO, Kales A, Rogan PK, Mascari M, Centurione A, Vela-Bueno A: Daytime sleepiness and REM abnormalities in PWS: Evidence of generalized hypoarousal. Int J Neurosci .*87*: 127-139, 1996.

44. Saitoh S, Buiting K, Cassidy SB, Conroy JM, Driscoll DJ, Gabriel JM, Gillessen-Kaesbach G. Glenn CC, Greenswag LR, Horsthemke B. Kondo I, Kuwajima K, Niikawa N. Rogan PK, Schwartz S, Seip J, Williams CA, Wiznitzer M, Nicholls RD: Clinical spectrum and molecular diagnosis of imprinting mutation patients: A new class of Prader-Willi and Angelman syndromes. Am J Med Genet. *68*: 195-206, 1997.

45. Butler MB, Rogan PK, Hedges LK, Cassidy SB, Moeschler JB: Kleinfelter and Trisomy X syndromes in patients with Prader-Willi syndrome and uniparental maternal disomy of chromosome 15 - A Coincidence? Am J Med Genet. *72*: 111-114, 1997.

46. Gocke C, Benko F, Rogan PK: Transmission of mitochondrial DNA heteroplasmy in normal pedigrees. Hum Genet. *102*: 182-186, 1998.

47. Rogan PK, Faux B, Schneider TD: Information analysis of human splice site mutations Hum Mutat. *12(3):* 153-171, 1998.

48. Allikmets R, Wasserman WW, Hutchinson A, Smallwood P, Nathans J, Rogan PK, Schneider D, Dean M: Organization of the ABCR gene: analysis of promoter and splice junction sequences Gene, *215*: 111-122, 1998.

49. Tooley PW, Salvo JJ, Schneider TD, Rogan PK: Phylogenetic inference using information theory-based PCR amplification. J Phytopathology, *146(8-9)*: 427-430, 1998.

50. Kannabiran C, Rogan PK, Basti S, Rao GN, Kaiser-Kupfer M, Hejtmancik JF: Autosomal dominant zonular cataract with sutural opacities is associated with a splice mutation in the βA3/A1-crystallin gene. Mol Vision, *4*: 21, 1998 http://www.molvis.org/molvis/v4/p21 .

51. O'Neill JP, Rogan PK, Cariello N, Nicklas JA: Mutations in the human HPRT gene which alter RNA splicing: A review of the spectrum. Mutat Res. 411(3):179-214, 1998.

52. Rogan PK, Seip JW, White L, Wenger SW, Menon R, Knoll JHM: Relaxation of imprinting in Prader-Willi syndrome. Hum Genet, 103(6): 694-701, 1998.

53. Ohta T, Gray TA, Rogan PK, Gabriel JM, Buiting K, Saitoh S, Methi M, Driscoll DJ, Horsthemke B, Butler MG, Nicholls RD: Imprinting mutation mechanisms in Prader-Willi syndrome. Am J Hum Genet *64*: 397-413, 1999.

54. Rogan PK, Sabol DW, Punnett H: Maternal uniparental disomy of chromosome 21 in a normal child. Am J Med Genet *83*: 69-71, 1999.

55. Martin RA, Sabol DW, Rogan PK: Maternal uniparental disomy of chromosome 14 confined to an interstitial segment (14q23-14q24.2). J Med Genet *36*: 633-636, 1999.

56. Kodolitsch Yv, Pyeritz RE, Rogan PK: Splice site mutations in atherosclerosis candidate genes: Relating individual information to phenotype. Circulation *100*: 693-699, 1999.

57. Amos-Landgraff JM, Ji Y, Gottlieb W, Depinet T, Wandstraat AE, Cassidy SB, Driscoll DJ, Rogan PK, Schwartz S, and Nicholls RD: Chromosome breakage in the Prader-Willi and Angelman Syndromes involves recombination between large, transcribed repeats at both breakpoints. Am J Hum Genet, *65*: 370-386, 1999.

58. Vockley J, Rogan PK, Anderson BD, Willard J, Seelan RS, Smith DI, and Liu W: An Unusually High Frequency of Abnormal Splicing of *IVD* RNA in Isovaleric Acidemia, Including Exon Skipping Caused by Missense Mutations in the *IVD* Gene. Am J Hum Genet, *66*:356-367, 2000.

59. Svojanovsky S, Schneider T, Rogan PK: Redundant designations of BRCA1 intron 11 splicing mutation. Hum. Mutation, 16: 264, 2000.

60. von Kodolitsch Y, Nienaber CA, Fliegner M, Rogan PK: Splice site mutations in atherosclerosis: mechanisms and predictive models. Z. Kardiol 90: 87-95, 2001.

61. Rogan PK, Cazcarro PM, Knoll JHM: Sequence-based design of single copy genomic DNA probes for fluorescence *in situ* hybridization, Genome Research, 11: 1086-1094, 2001.

62. Thompson TE, Rogan PK, Risinger JI, Taylor JA: Splice Variants, But Not Mutations, of DNA Polymerase β Are Common in Bladder Cancers, Cancer Research, 62: 3251-3256, 2002.

63. Rogan PK, Svojanovsky SR, Leeder JS: Information theory-based analysis of CYP219, CYP2D6 and CYP3A5 splicing mutations, Pharmacogenetics, 13(4): 207-218, 2003.

64. Knoll JHM, Rogan PK. Sequence-based, *in situ* detection of chromosomal abnormalities at high resolution, American Journal of Medical Genetics, 121A: 245-257, 2003.

65. Consevage M, Kasarda S, Sabol D, Rogan PK: Genetic mapping of familial hypertrophic-restrictive cardiomyopathy, Hum Genet., in press.

66. Gadiraju S, Vyhlidal CA, Leeder JS, Rogan PK: Genome-wide prediction, display and refinement of binding sites with information theory-based models, BMC Bioinformatics, 4:38 (http://www.biomedcentral.com/1471-2105/4/38).

67. von Kodolitsch Y, Berger J, Rogan PK: Predicting Severity of Hemophilia A and B Splicing Mutations by Information Analysis, Human Mutation, submitted.

68. Lamba V, Lamba J, Stromb S, Davilac J, Hancocka M, Yang W, Fackenthald JD, Rogan PK, Ring B, Wrighton S, Schuetz EG. Hepatic CYP2B6 Expression: Gender and Population Differences and Relationship to CYP2B6 Genotype and CAR Expression. J Pharmacol Exp Ther. 2003 Dec; 307(3): 906-22.

69. Knoll JHM and Rogan PK. High Resolution Definition of Chromosome Abnormalities with Probes Designed from Genome Sequences. Discovery Medicine, 2004, 4 (21): 99-101.

70. Bi C and Rogan PK Bipartite Pattern Discovery by Entropy Minimization of Unaligned DNA Sequences, 2004, Nucl. Acids Res, 32:4979-4991.

71. Vyhlidal C, Rogan PK and Leeder JS. Development and Refinement of Pregnane X Receptor DNA Binding Site Model Using Information Theory, J. Biol. Chem., 2004, doi:10.1074/jbc.M408395200.

BOOK CHAPTERS, LETTERS AND CONFERENCE REPORTS

1. Nicholls RD, Gottlieb W. Mascari MJ, Rinchik EM, Pai GS, Driscoll DJ, Butler MG, Zori RT, Neumann PE, Waters MF, Zackowski JL, Horsthemke B. Rogan PK, Ladda RL, Williams CA: Molecular Analysis in Angelman Syndrome, Prader-Willi Syndrome and Potential Mouse Models. In: NATO Advanced Research Workshop on Prader-Willi Syndrome and Other Chromosome 15q Deletion Disorders. S.B. Cassidy (ed). Springer-Verlag, Berlin, 1992.

2. Rogan PK, Salvo JJ: High fidelity amplification of ribosomal gene sequences from mummified South American human remains. In: Ancient DNA, B. Herrmann (ed). Springer-Verlag, New York, pp. 182-194, 1993.

3. Rogan PK, Salvo JJ: High fidelity polymerase chain reaction amplification products from mummified South American human remains. In: Proceedings of the First World Congress on Mummy Studies, C Rodriguez-Martin (ed). Museo Archeologia y Etnografica: Tenerife, 1995.

4. Robinson WP, Horsthemke B, Leonard S, Malcolm S, Morton C, Nicholls RD, Ritchie R, Rogan P, Schultz R, Schwartz S, Sharp J, Trent R, Wevrick R, Williamson M, Knoll JHM: Report of the third international chromosome 15 mapping workshop, Vancouver Canada 1996. Cytogenet Cell Genet. 1997;76(1-2):1-13.

5. Rogan PK: Masked mosaicism, J. NIH Research, *9*: 17-18, 1997

6. Knoll JHM, Rogan PK: Prader Willi Syndrome, Encyclopedia of Neuroscience, 3rd edition (George Adelman and Barry H. Smith, eds.), 2004.

7. Rogan PK, Knoll JHM: High Resolution Detection of Chromosome Abnormalities with Single Copy Fluorescence in situ Hybridization. in Proceedings of the 2004 IEEE International Symposium on Biomedical Imaging: From Nano to Macro, Arlington, VA, USA, 15-18 April 2004. IEEE 2004, pp 73-76

8. Bi C, Vyhlidhal C, Leeder JS, Rogan PK: A minimization entropy based bipartite algorithm and its application to PXR/RXRα binding sites. *RECOMB 2004* 453-454 (San Diego, CA USA, 2004).

9. Kobayashi T, He L, Shyu C-R, Knoll JHM, Rogan PK: Content and Classification based Ranking Algorithm for Metaphase Chromosome Images, IEEE Conference on Multimedia Imaging, 2004.

10. Bi C and Rogan PK: Information theory as a model of genomic sequences, Encyclopedia of Bioinformatics, Genomics and Proteomics, Wiley NY, in press.

ABSTRACTS

1. Wilson SR, Corvan PJ, Seiders RP, Hodgson DJ, Brookhart M, Hatfield WE, Miller JS, Reiss AH, Rogan PK, Gebert E, Epstein AJ : « The Structure and Magnetic and Electrical Conductivity Properties of the Charge Transfer Compound 1,1-Dimethylferrocenium Bis(tetracyanoquinodimethane), $(CH_3C_5H_3)_2Fe(TCNQ)_2$," NATO Advanced Research Institute on Molecular Metals, Les Arcs, France, 10-16 Sept 1978 [Paper P-46].

2. Salvo JJ, Allison MJ, Rogan PK: Molecular genetics of pre-Columbian South American mummies. Am J Phys Anthr 78(2):295, 1989.

3. Rogan PK, Salvo JJ: Molecular genetics of pre-Columbian South American mummies. J Cell Biochem 13C:123, 1989.

4. Salvo JJ, Rogan PK: Identification and amplification of DNA sequences in pre-Columbian South American mummies. Sixth Conversation in Biomolecular Stereodynamics, Albany, NY, 1989.

5. Rogan PK, Salvo JJ, Tooley PW: Use of universal PCR primers to amplify 28S ribosomal DNA from taxonomically diverse organisms. Fourth International Congress of Systematic and Evolutionary Biology, College Park, Maryland, 1990.

6. Rogan PK, Klar A, Strathern JN, Lemkin P: Identification of cell type specific chromosomal loci by in vivo methylation. Symposium of the International Electrophoresis Society, 1991.

7. Mascari M, Rogan P. Ladda R. Butler M, Gottlieb W. Nicholls R: Molecular diagnosis of Prader-Willi Syndrome. 8th International Congress of Human Genetics, 1991.

8. Rogan P. Mascari M, Ladda R. Gottlieb W. Nicholls R: The origin of maternal disomy in Prader-Willi Syndrome. 8th International Congress of Human Genetics, 1991.

9. Mowrey PN, Chorney M, Lerman M, Zbar B. Latif F. Rogan PK, Ramer J. Ladda R. Further molecular analysis of the deletion of 3p25 syndrome. 8th International Congress of Human Genetics, 1991.

10. Venditti CP, Rogan PK, Chorney MJ: Alumorph PCR analysis of MHC Class I clones. Eighth International H-2/HLA Workshop, 1992.

11. Rogan P. Salvo JJ: High-fidelity polymerase chain reaction amplification products from mummified South American Human remains. First International Congress on Mummy Studies, 1992.

12. Nicholls RD, Gottlieb W. Avidano K, Jong M, Driscoll DJ, Mascari MJ, Rogan PK, Horsthemke B. Russell LB, Rinchik EM: Mammalian genomic imprinting: Prader-Willi and Angelman syndromes and mouse models. NIH Conference on Genomic Imprinting. Bethesda, MD, April 12-13, 1992.

13. Rogan PK, Mascari MJ, Ladda RL, Nicholls RD: Genetic mapping with Prader-Willi patients carrying recombinant disomic chromosomes. First International Workshop on Human Chromosome 15, Tucson, AZ, June 18-19, 1992.

14. Nicholls RD, Driscoll DJ, Rogan PK, Spritz RA, Gottlieb W. Jong M, Avidano KM, Waters MF, Glenn CC, Williams CA, Zori RT, Horsthemke B, Robinson W, Schinzel A, Saitoh S, Niikawa N, Russell LB, Bultman SJ, Rinchik EM: Mapping of loci in human chromosome 15qll-ql3 and a region of conserved synteny in mouse chromosome 7, including a candidate imprinted gene (D15S9) and the pink-eyed dilution (p/D15S12) gene. First International Workshop on Human Chromosome 15, Tucson, AZ, June 18-19, 1992.

15. Nicholls RD, Gottlieb W. Avidano KM, Jong MTC, Horsthemke B. Russell LB, Bultman SJ, Spritz RA, Rogan PK, Rinchik EM: Mouse models for genomic imprinting and phenotypic features in Prader-Willi and Angelman syndromes. Prader-Willi Syndrome Association (USA) 14th Annual Conference, Philadelphia, PA, July 15, 1992.

16. Rogan PK, Mascari MJ, Nicholls RD, Ladda RL: Development of a molecular genetic database for patients with Prader-Willi syndrome. 14th Annual Conference of the Prader-Willi Syndrome Association, July 15, 1992.

17. Mascari MJ, Rogan PK, Gannutz LS, McCurdy MP, Croft CD, Lichty TR, Ladda RL: Pseudomosaicism Trisomy 15 in amniocytes: Concern about the possibility of uniparental disomy in the fetus. 14th Annual Conference of the Prader-Willi Syndrome Association, July 15, 1992.

18. Rogan PK, Mascari MJ, Ladda RL: Genetic mapping with Prader-Willi patients carrying recombinant disomic chromosomes. Clinical Research, Eastern Meeting, New York, NY, October 9-10, 1992.

19. Rogan PK, Mascari MJ, Ladda RL, Nicholls RD: Genetic mapping with Prader-Willi patients carrying recombinant disomic chromosomes. American Society of Human Genetics, San Francisco, CA, November 9-15, 1992.

20. Salvo JS, Aufderheide AC, Rogan PK: Kinship studies in ancient human populations. American Society of Human Genetics, San Francisco, CA, November 9-15, 1992.

21. Floros J. Kotikalapudi P. Rogan P: Localization of genetic variability in human surfactant protein B gene and its association with respiratory distress syndrome. American Society of Human Genetics, San Francisco, CA, November 9-15, 1992.

22. Rogan PK, Salvo JS, Aufderheide AC: Kinship studies in ancient human populations. 3rd International Congress on Human Paleontology, Jerusalem, Israel, August 1993.

23. Butler MG, Driscoll DJ, Papenhausen PR, Johnson VP, Rogan PK: Analysis of 15q25Dqter markers in patients with ring 15 and Russell-Silver syndromes. American Society of Human Genetics, 1993.

24. Gottlieb W. Rogan PK, Ledbetter DH, Driscoll DJ, Nicholls RD: Analysis of chromosome-breakage mechanisms in Prader-Willi and Angelman syndromes. American Society of Human Genetics, 1993.

25. Rogan PK, Lichty TR, Ladda RL, Mascari MJ, Steele MW, Wenger SL, Malcolm S. Driscoll DJ, Nicholls RD: Uniparental disomy in Angelman Syndrome: A consequence of paternal meiotic non-disjunction. American Society of Human Genetics, 1993.

26. Floros J. Rishi A, Veletza SV, Rogan PK: Concerted and independent genetic events in the 3'untranslated region of the human surfactant protein A genes. American Society of Human Genetics, 1993.

27. Woodage T. Prasad M, Dixon JW, Selby RE, Romain DR, Columbano-Green LM, Graham D, Rogan P. Smith A, Trent RJ: Bloom syndrome and maternal uniparental disomy for chromosome 15. American Society of Human Genetics, 1993.

28. Guida L, Rogan PK, Chakravarti A, Ledbetter DH, Schwartz S. Nicholls RD: Isolation of $(CA)_n$ repeats and characterization of the pericentromeric region of chromosome 15q. American Society of Human Genetics, 1993.

29. Mascari MJ, Ladda RL, Woodage T. Trent RJ, Lai LW, Erickson RP, Cassidy SB, Petersen MB, Mikkelsen M, Driscoll DJ, Nicholls RD, Rogan PK: Perturbed recombination of chromosome 15 in Prader-Willi patients with maternal disomy. American Society of Human Genetics, 1993.

30. Phillips RL, Rogan PK, Culiat B. Stubbs L, Rinchik EM, Gottlieb W. Nicholls RD: A YAC contig spanning 4 genes in distal human chromosome 15qll-ql3, mapping of the human GABRa3 gene and effect of homozygous deletion of three GABAA receptor genes in mouse. American Society of Human Genetics, 1993.

31. Lentz SE, Rogan PK, Martin CR: Mitochondrial DNA studies of the origins of the Aboriginal people of the Canary Islands. Second International Ancient DNA Conference, Washington, DC, 1993.

32. Rogan PK, Lentz SE: Molecular genetic evidence suggesting treponematosis in pre-Columbian, Chilean mummies. American Association of Physical Anthropologists, 1994.

33. Lentz SE, Rogan PK, Martin CR: Mitochondrial DNA studies of the origins of the Aboriginal people of the Canary Islands. American Association of Physical Anthropologists, 1994.

34. Woodage T. Prasad M, Dixon JW, Selby RE, Romain DR, Columbano-Green LM, Graham D, Seip JR, Rogan PK, Smith A, Trent RJ: Localization of the Bloom syndrome gene by homozygosity mapping in a patient with maternal uniparental disomy of chromosome 15. Chromosome 15 International Workshop, Oxford, U.K.

35. Rogan PK, Mascari MJ, Ladda RL, Woodage T. Trent RJ, Smith A, Lai LW, Erickson RP, Cassidy SB, Petersen MB, Mikkelsen M, Driscoll DJ, Nicholls RD: Perturbed recombination of chromosome 15 in Prader-Willi patients with maternal disomy. Chromosome 15 International Workshop, Oxford, U.K.

36. Butler MG, Woodward AL, Driscoll DJ, Papenhausen PR, Johnson VP, Raskin S. Rogan PK: Molecular genetic analysis of 15q26.1-qter markers in patients with ring 15 and Russell-Silver syndromes. Chromosome 15 International Workshop, Oxford, U.K.

37. Consevage MW, Salada GC, Baylen BG, Ladda RL, Rogan PK: Identification of a new missense mutation in the b-cardiac heavy chain myosin gene in patients with familial hypertrophic cardiomyopathy. Society for Pediatric Research, 1994.

38. Ladda RL, Close P. Gannutz LS, Seip JR, Antonarakis SE, Rogan PK: Acute monoblastic leukemia in Trisomy 21: Tetrasomy 21 in the monoblast clone. American College of Medical Genetics, 1994.

39. Ramer JC, Rogan PK, Mascari MJ, Ladda RL: Brothers with findings common to Bardet-Biedl syndrome and Albright's hereditary osteodystrophy. American College of Medical Genetics, 1994.

40. Rogan PK, Mascari MJ, Ladda RL, Woodage T. Trent RJ, Smith A, Lai LW, Erickson RP, Cassidy SB, Petersen MB, Mikkelsen M, Driscoll DJ, Nicholls RD, Butler MG: Coinheritance of other chromsome 15 abnormalities with Prader-Willi syndrome: Genetic risk estimation and mapping. Prader-Willi Syndrome Association Scientific Conference, 1994.

41. Saitoh S. Seip J. Tommerup N. Greenswag L, Rogan PK, Nicholls RD: The imprinted SNRPN gene is associated with a polycistronic mRNA and an imprinting control element. American Society of Human Genetics, 1994.

42. Amos-Landgraf J. Gottlieb W. Rogan PK, Nicholls RD: Chromosome breakage in Prader-Willi and Angelman Syndrome deletions may involve recombination between a repeat at the proximal and distal breakpoints. Am Soc Hum Genet, 1994.

43. Rogan PK, Close P. Blouin JL, Seip JR, Gannutz LS, Ladda RL, Antonarakis SE: Duplication and loss of chromosome 21 in two children with Down Syndrome and acute leukemia. American Society of Human Genetics, 1994.

44. Gocke CD, Dodson WC, Benko FA, Seip JR, Rogan PK: Patterns of human mitochondrial DNA transmission in oocytes and extended families. American Society of Human Genetics, 1994.

45. Belchis DA, Meece CA, Rogan PK, Williams R. Gocke CD: Detection of molecular changes at the retinoblastoma locus in osteosarcomas by the polymerase chain reaction. Annual Meeting of the United States and Canadian Academy of Pathology, March 11-17, 1995.

46. Rogan PK, Seip JR, Helm K, Miller B: Chloroma in a child with previous acute myelogenous leukemia. Middle Atlantic Regional Human Genetics Network Cytogenetics Workshop, September 20, 1994, Philadelphia, PA.

47. Consevage MW, Seip JR, Belchis DA, Davis AT, Baylen BG, Rogan PK: Novel association of a mosaic chromosomal 22qll deletion associated with hypoplastic left heart syndrome. American Heart Association Scientific Conference on the Molecular, Cellular and Functional Aspects of Cardiovascular Development, 1995.

48. Amos-Landgraf J, Gottlieb W, Rogan PK, Schwartz S, Cassidy S, Nicholls RD: Chromosome breakage in Prader-Willi and Angelman syndrome deletions involves recombination between repeats at both breakpoints. Great Lakes Regional Genetics Group, March 24-26, 1995, Columbus, OH.

49. Rogan PK, Seip JR, Saitoh S. Nicholls RD: Long-range, parental-specific gene expression in Prader-Willi syndrome is modulated by an imprinting control element. Fourteenth Summer Symposium in Molecular Biology, August 3-5, 1995, State College, PA.

50. Rogan PK, Mascari MJ, Ladda RL, Woodage T. Trent RJ, Smith A, Lai LW, Erickson RP, Cassidy SB, Petersen MB, Mikkelsen M, Driscoll DJ, Nicholls RD, Butler MG: Increased risk of coinheritance of atypical findings in patients with Prader-Willi syndrome due to maternal uniparental disomy, Middle Atlantic Regional Human Genetics Network Molecular and Cytogenetics Workshop, September 13, 1995, Philadelphia, PA.

51. Knoll JHM, Rogan PK, Nicholls RD, Wu B, Korf B, White L: Allele-specific replication of 15qllql3 loci: A diagnostic assay for uniparental disomy, American Society of Human Genetics, October, 1995.

52. Gocke CD, Simmons Z, Benko FA, Towfighi J, Rogan PK: Chronic progressive external ophthalmoplegia (CPEO) and "limb-girdle dystrophy" in two members of a family with different mitochondrial DNA (mtDNA) deletions. American Society of Human Genetics, October, 1995.

53. Saitoh S, Rogan PK, Buiting K, Schwartz S, Cassidy SB, Glenn CC, Driscoll DJ, Horsthemke B, Nicholls RD: Minimal definition of the imprinting center and fixation of a chromosome 15qll-ql3 epigenotype by imprinting mutations. American Society of Human Genetics, October, 1995.

54. Rogan PK, Seip JR, Knoll JHM, White LS, Wenger SL, Steele MR, Sperling M, Aparicio L, Menon R: Relaxation of imprinting in patients with Prader-Willi Syndrome. 11th Annual Prader Willi Syndrome Scientific Conference. July, 1996.

55. Ohta T, Buiting K, Gabriel JM, Schwartz S, Cassidy SB, Rogan PK, Glenn CC, Driscoll DJ, Horsthemke B, Nicholls RD: Imprinting mutations in Prader-Willi and Angelman Syndrome exemplify a new genetic mechanism. 11th Annual Prader Willi Syndrome Scientific Conference. July, 1996.

56. Rogan PK, Butler MG: Atypical clinical findings in PWS patients. 11th Annual Prader Willi Syndrome Scientific Conference. July, 1996.

57. Rogan PK, Seip JR, Knoll JHM, White LS, Wenger SL, Steele MR, Sperling M, Aparicio L, Menon R: Relaxation of imprinting in patients with Prader-Willi Syndrome. American Society of Human Genetics, October, 1996.

58. Faux BM, Schneider TD, Rogan PK: Information analysis of human splice recognition site mutations. American Society of Human Genetics, October, 1996.

59. Ohta T, Rogan PK, Saitoh S, Buiting K, Horsthemki B, Weksberg R, Ishikawa T, Butler MG, Gabriel JM, Nicholls RD: Minimal definitition and function of the imprinting center for Prader-Willi syndrome. American Society of Human Genetics, October, 1996.

60. Ohta T, Saitoh S, Buiting K, Rogan PK, Butler MG, Deng G, Driscoll DJ, Gabriel JM, Malandro MS, Gray TA, Dittrich B, Horsthemke B, Nicholls RD: Minimal definition and function of the PWS/AS imprinting center in germline switching of the imprint over a 2 Mb domain in chromosome 15q11-q13. Third International Workshop on Human Chromosome 15, October 1996.

61. Rogan PK, Mascari MJ, Ladda RL, Woodage T, Trent RJ, Smith A, Lai LW, Erickson RP, Cassidy SB, Petersen MB, Mikkelsen M, Driscoll DJ, Nicholls RD, Butler MG: Coinheritance of other chromosome 15 abnormalities with Prader-Willi syndrome: Genetic risk estimation and mapping. Third International Workshop on Human Chromosome 15, October 1996.

62. Rogan PK, Faux BM, Schneider TD: Information analysis of human splice junction mutations. HUGO-Mutation Database Association, October 1996.

63. Rogan PK, Faux BM, Schneider TD: Information analysis of human splice junction mutations. Annual NIH Research Symposium, November 22-23, 1996.

64. Rogan PK, Schneider TD, Faux B, Wijnen J, Radice P, Baba S, Scott R, Viel A, Genuardi M, Meera Khan P, Fodde R, and the ICG-HNPCC: Information theory-based analysis of splice junction mutations in hereditary non-polyposis colon cancer (HNPCC), American Society of Human Genetics, October 1997.

65. Rogan PK, Faux BM, Schneider TD: Information analysis of human splice junction mutations. TIGR Conference on Computational Genomics, November 1997.

66. Rogan PK: Molecular diagnosis using information theory-based methods. American Association for the Advancement of Science Annual Meeting, February 1998.

67. Rogan PK, Schneider TD, Faux B, Wijnen J, Radice P, Baba S, Scott R, Viel A, Genuardi M, Meera Khan P, Fodde R, and the ICG-HNPCC: Information theory-based analysis of splice junction mutations in hereditary non-polyposis colon cancer (HNPCC), American Association of Cancer Research, March 1998.

68. Martin R, Sabol D, Rogan PK: Segmental uniparental maternal disomy of chromosome 14. David Smith Clinical Genetics Workshop, August 1998.

69. Rogan PK, Schneider TD: Comparison of algorithms for computational identification of natural and mutant human splice sites. HUGO-Mutation Database Association, October 1998.

70. Rogan PK, Sabol DW, Close P: Localization of genetic loci that predispose to acute leukemia in patients with Down syndrome, American Association for Cancer Research, April 1999.

71. Gocke CD, Benko FA, Dodson W, Rogan PK: Mitochondrial heteroplasmy in normal human ova and sperm,_Serono Symposia Australasia International Symposium: "The Bottleneck: Gamete and Embryo Mitochondria in Humans," May 1999.

72. Punnett HH, Schneider AS, Sabol DW, Bardakjian TM, Martin KA, Howard-Peebles PN, Rogan PK: Predominance of Down syndrome phenotype in mos 45,X,+mar(X)[18]/46,XX,+21,der(21;21)(q10;q10)[2] in cardiac tissues, American Society of Human Genetics, October 1999.

73. Rogan PK, Sabol DW, Close P: Localization of genes predisposing to acute leukemia in Down syndrome, American Society of Human Genetics, October 1999.

74. Gocke CD, Benko FA, Dodson W, Rogan PK: Normal human sperm exhibit greater mitochondrial DNA (mtDNA) heteroplasmy than ova, American Society of Human Genetics, October 1999.

75. Consevage MW, Kasarda S, Sabol DW, Rogan PK: Genetic mapping of familial hypertrophic-restrictive cardiomyopathy, American Society of Human Genetics, October 1999.

76. Butler MG, Eskew JD, Wilcox LD, Rogan PK: X chromosome inactivation in families with Prader-Willi syndrome, American Society of Human Genetics, October 1999.

77. Knoll JHM, Cazcarro P, Rogan PK: Clinical application of sequence-based single copy probes for FISH, American Society of Human Genetics, October 2000.

78. Rogan PK, Cazcarro P, Knoll JHM. Single copy hybridization probes derived by genomic sequence analysis. American Society of Human Genetics, October 2000.

79. Thompson TE, Rogan PK, Risinger JI, Taylor JA: Alternative splicing of DNA Polymerase β mRNA in normal tissues and bladder cancer. American Society of Human Genetics, October 2000.

80. Kodolitsch Yv, Nienaber CA, Schalwat I, Svojanovsky SR, Rogan PK: Relationship of splice site information to protein activity in hemophilia A and B. American Society of Human Genetics, October 2000.

81. Svojanovsky SR, Rogan PK: Tumor suppressor function of the BRCA1 gene has a low threshold for mutation. American Society of Human Genetics, October 2000.

82. Rogan PK, Cazcarro P, Knoll JHM: Single copy hybridization probes for detection of chromosome rearrangements derived by genomic sequence analysis. American Association for Cancer Research, Proceed Am Assoc Ca Res 42:336. March 2001.

83. Rogan PK, Cazcarro P, Knoll JHM: Single copy hybridization probes for detection of chromosome rearrangements derived by genomic sequence analysis. 10th International Congress of Human Genetics (platform), May 2001.

84. Hurwitz I*, Svojanovsky S, Rogan PK, Leeder JS: Modulation Of Constitutive Cyp2d6 Activity: Differential Binding Of Nf-κB P50 To The –1496C And –1496G Variants. 6th Annual Meeting of the International Society for Study of Xenobiotics, Drug Metab Rev 33(Suppl1):95:188., Munich Germany, October 2001.

85. Svojanovsky SR, Hobson GM, Sperle K, Sistermans EA, Garbern JY, Rogan PK: Predicted expression of PLP splicing mutations in Pelizaeus-Merzbacher disease. American Society of Human Genetics, October 2001.

86. Rogan PK, Cazcarro P, Knoll JHM: Novel features of genome organization revealed by single copy FISH. American Society of Human Genetics, October 2001.

87. Cazcarro P, Rogan PK, Knoll JHM: Rapid sequence-based definition of chromosomal abnormalities. American Society of Human Genetics, October 2001.

88. Hurwitz I, Svojanovsky S, Leeder JS, Rogan PK: Modeling differential binding of NF-kB p50 to a CYP2D6 promoter variant by information theory. American Society of Human Genetics, October 2001.

89. J. Tazelaar J, Uhrmacher J, Svojanovsky S, Rubinstein W, Myers E, Rogan P, Baysal B, Kant J: Hereditary Paraganglioma: Validation and Early Diagnostic Experience. Association for Molecular Pathology, November 2001 (published in J. Molecular Diagnostics 2001).

90. Rogan PK, Leeder JS: Modeling splice site and transcription factor binding site variation by information theory. Keystone Symposium on Molecular and Cellular Biology: Genotype to Phenotype-Focus on Disease (invited), February 2002.

91. Cambi F, Rogan PK, Svojanovsky S, Hobson G: Correlation of genotype and phenotype in Pelizaeus-Merzbacher disease. American Neurology Association, 2002.

92. Knoll JHM, Rogan PK: Detection of chromosomal rearrangements with sequence-defined, single copy hybridization probes. Second International NCI-EORTC Meeting on Cancer Diagnostics: From Discovery to Clinical Practice. June, 2002.

93. Rogan PK, Svojanovsky S, Hurwitz I, Schneider TD, Leeder JS: Modeling splice site and transcription factor binding site variation by information theory. American Society of Human Genetics, October 2002.

94. Vyhlidal CA, Rogan PK, Leeder JS: Modeling PXR/RXRα Binding Using Information Theory. 7th Annual Meeting of the International Society for Study of Xenobiotics, October 2002.

95. Rao V, Rogan PK: Localization of genes predisposing to acute leukemia in Down syndrome-A Childrens' Oncology Group study. American Society of Human Genetics, October 2002.

96. Knoll JHM, Angell P, Rogan PK: Detection of chromosomal rearrangements with single copy FISH probe arrays. American Society of Human Genetics, October 2002.

97. Gaedigk A, Abdel-Rahman SM, Marcucci, KA, Pearce, RE, Rogan PK, Bradford LD, Spielberg SP, Leeder, JS. Characterization of an African American (AA) subject carrying two novel functional CYP2D6 alleles. American Society of Clinical Pharmacology and Therapeutics, April 2 2003 [PI-40].

98. Gadiraju S, Rogan PK. Computational protein binding site analysis in complete genomes. 2nd Annual Kansas City Area Life Sciences Day, March 2003.

99. Gaedigk A, Abdel-Rahman SM, Marcucci KA, Pearce R, Rogan PK, Bradford LD, Leeder JS. Characterization of an African American (AA) subject carrying two novel functional CYP2D6 alleles. 2nd Annual Kansas City Area Life Sciences Day, March 2003.

100. Vyhidal CA, Rogan PK, Leeder JS. Binding & Modeling PXR/RXR Using Information Theory. 2nd Annual Kansas City Area Life Sciences Day, March 2003.

101. Rogan PK, Vylidal CA, Svojanovsky SR, Leeder JS. Modeling Splice Site and Transcription Factor Binding Site Variation by Information Theory. 2nd Annual Kansas City Area Life Sciences Day, March 2003

102. Knoll JHM, Rogan PK. Detection of Chromosomal Rearrangements with Single Copy FISH. 2nd Annual Kansas City Area Life Sciences Day, March 2003.

103. Rogan PK Rao VN. Localization of genes predisposing to acute leukemia in Down syndrome-A Children's Oncology Group Study. 2nd Annual Kansas City Area Life Sciences Day, March 2003.

104. Rao, VN, Rogan, PK. Localization of genes predisposing to acute leukemia in Down syndrome-A Children's Oncology Group Study. American Association for Cancer Research, accepted for platform presentation.

105. V. Rao and P.K. Rogan. Localization of genes predisposing to acute leukemia in Down syndrome-A Childrens Oncology Group Study. XIX International Congress of Genetics, July 2003.

106. P.K. Rogan, C.A. Vyhidal and J.S. Leeder. Binding & Modeling PXR/RXRα Using Information Theory. XIX International Congress of Genetics, July 2003

107. J.H.M. Knoll & P.K. Rogan. Detection of Chromosomal Rearrangements with Single Copy FISH. XIX International Congress of Genetics, July 2003.

108. R. Gaedigk, A. Gaedigk, K. Marcucci, R. Pearce, P.K. Rogan and J.S. Leeder. Developmental Expression Of Cytochrome P450 3A4, 3A5, 3A7 and 3A43 And Characterization Of CYP3A5 Splice Variants. Joint Welcome Trust/Cold Spring Harbor Laboratory Meeting on Pharmacogenomics, September 2003.

109. Will Marrying Pharmacoepidemiology And Pharmacogenetics Produce The Rosetta Stone For Collaborative Investigations Of Drug-Induced Birth Defects? Leeder, J.S.[1], Gaedigk, A.[1], Gaedigk, R.[1], Louik, C.[2], Rogan, P.K.[1], Werler, M.[2] and Mitchell, A.A. Joint Welcome Trust/Cold Spring Harbor Laboratory Meeting on Pharmacogenomics, September 2003.

110. Gaedigk A, Gaedigk R, Rogan PK, and Leeder JS. Extensive alternative splicing of cytochrome P4502D6 (CYP2D6) mRNA: explanation for variability among subjects with identical genotypes? American Society of Clinical Pharmacology and Therapeutics, March 2004.

111. Mora JR, Knoll JHM, Rogan PK, Wilson GS, Getts RC. Enhanced detection of t(15;17) in Acute Promyelocytic Leukemia with single-copy FISH probes labeled with DNA dendrimers. Pittcon March 3-7, 2004, Chicago Ill.

112. P.K. Rogan, C.P. Bi, C.A. Vyhidal, S.Gadiraju, D. Dinakarpandian, and J.S. Leeder. Prediction and Validation of PXR/RXRα Binding Sites in Transcriptionally-responsive Genes. Keystone Symposium: Human Genome Sequence Variation and the Inherited Basis of Common Disease, January 8 - 13, 2004, Breckenridge Co.

113. C.P. Bi, C.A. Vyhidal, and J.S. Leeder, P.K. Rogan. Relating transcriptional enhancer strengths of PXR binding sites to information content. International Society of Computational Biology Regional Meeting, Dec. 4-6, 2003, Aspen Co.

114. Rogan PK: Analysis of DNA sequence variation by information theory-based analysis. 7th International Int. Soc. for the Study of Xenobiotics Meeting, Vancouver, Canada, August 2004, (Invited speaker)

115. Bi, CP, Vyhlidal CA, Leeder JS, and Rogan PK: A bipartite model of PXR/RXR binding sites based on information theory. 7th International Int. Soc. for the Study of Xenobiotics Meeting, Vancouver, Canada, August 2004.

116. Knoll JHM, Marion AM, Flejter W, Persons D, Cowan J, Rogan PK. Small deletions upstream of ABL1 are rare in CML. Proceedings of the 94th Annual Meeting of the AACR, April 2004.

117. D. Dinakarpandian, V. Raheja, S. Mehta, P.K. Rogan: Identification of Xenobiotic-responsive Target genes by Tandem Machine Learning. American Society of Human Genetics Annual Meeting, October 2004.

118. Knoll JHM, Marion AM, Fletjer W, Persons D, Cowan J, Rogan PK. Small upstream deletions of *ABL* 1 are rare in Chronic Myelogenous Leukemia. American Society of Human Genetics Annual Meeting, October 2004.

119. Knoll JHM, Rogan PK. Applications of personalized molecular cytogenetic diagnostics. American Society of Human Genetics Annual Meeting, October 2004.

120. C. Bi, C.Vyhlidal, J.S. Leeder, P.K. Rogan. A Minimization Entropy-Based Bipartite Algorithm with Application to PXR/RXRalpha Binding Sites. American Society of Human Genetics Annual Meeting, October 2004.

121. C.A. West, C. Bi, and P. K. Rogan. Identification of PXR/RXRα Transcriptional Targets by *in vitro* Competition with DNA Binding Sites Predicted by Information Theory. American Society of Human Genetics Annual Meeting, October 2004.

122. VK. Nalla and PK Rogan. Automated Information Theory-Based Mutation Analysis: Application to mRNA splicing. American Society of Human Genetics Annual Meeting, October 2004.

123. Yanala P, Lu T, El-Ghussein F, Zhao C, Medhi D, Wang Y-P, Knopp J, Knoll JHM, Rogan PK. Automated detection of metaphase chromosomes in FISH and routine cytogenetics. American Society of Human Genetics Annual Meeting, October 2004.

124. VK. Nalla and PK Rogan. Automated Information Theory-Based Mutation Analysis: Application to mRNA splicing. Human Genome Variation Society, October 2004.

Joan H.M. Knoll, Ph.D., FACMG, FCCMG
Children's Mercy Hospital and Clinics
2401 Gillham Road
Kansas City, MO 64108
Tel: (816) 983-6509
Fax: (816) 983-6515
E-mail: jknoll@cmh.edu

Education:

1979 B.Sc. (Biology, Genetics) 1979, University of Western Ontario
1982 M.Sc. (Genetics) 1982, University of Saskatchewan, Saskatoon
1987 Ph.D. (Human Genetics) 1987, University of Manitoba

Postdoctoral Training:

Research and Clinical Genetics Laboratory Fellowship:

1987-90 Research Fellow in Medicine (Genetics), Children's Hospital, Boston
1987-90 Fellow in Pediatrics, Harvard Medical School

Certification:

1990 Board Certified in Clinical Cytogenetics, American Board of Medical Genetics
1992 Board Certified in Clinical Cytogenetics, Canadian College of Medical Genetics
1992 Fellow, Canadian College of Medical Genetics
1992 Founding Fellow in American College of Medical Genetics
1996 Board Certified in Clinical Molecular Genetics, American College of Medical Genetics

Academic Appointments:

1990 Instructor in Pediatrics, Harvard Medical School
1991 Instructor in Pathology, Harvard Medical School
1992-99 Assistant Professor in Pathology, Harvard Medical School
1999- Associate Professor in Medicine/Pediatrics, UMKC School of Medicine

Hospital and Other Appointments:

1990-99 Research Associate in Medicine (Genetics), Children's Hospital, Boston
1990-91 Consultant, Integrated Genetics, Framingham, MA
1991-98 Co-director, Clinical Cytogenetics Service, Beth Israel Deaconess Medical Center, Boston
1998-99 Director, Clinical Cytogenetics Service, Beth Israel Deaconess Medical Center, Boston
1999- Director, Clinical Cytogenetics Service, Children's Mercy Hospital, Kansas City
1999- Director, Human Genetics Research Laboratory, Children's Mercy Hospital, Kansas City
1999- Professorship in Pediatric Cytogenetics, Children's Mercy Hospital, Kansas City

Committee Assignments:

National and Regional:

1991-1995 Scientific Advisor, Angelman Syndrome Foundation
1992- Scientific Advisor, Canadian Angelman Syndrome Society
1994- Scientific Advisor, Scientific Advisor, Inverted Duplication 15 Education and Advocacy Group
1994- Member, Technology Transfer Subcommittee for diagnostic testing of Angelman and Prader-Willi syndromes, American College of Medical Genetics
1996- Co-organizer of Third International Chromosome 15 Workshop in Vancouver, BC, October 1996
1999- Member, UM Columbia/Children's Mercy Hospital Genetics Fellowship Joint Training Program

2001	Judge, Fellows, Residents & Students Research Presentations, Children's Mercy Hospital's Research Days, June 2001
2002-03	Advising Member, Johnson County Community College Biotechnology Steering Committee, Overland Park, KS
2003-	Member, Basic Sciences Research Committee, Children's Mercy Hospitals, Kansas City

Memberships and Committee Assignments in Professional Societies:

1986-	American Society of Human Genetics
1989-	Reviewer for Genetics journals (American Journal of Human Genetics, American Journal of Medical Genetics, Cancer Genetics and Cytogenetics, Cytogenetics and Cell Genetics, Genomics, Human Molecular Genetics, New England Journal of Medicine)
1991-97	Member, Board of Directors, Angelman Syndrome Foundation
1992-	Fellow in Canadian College of Medical Genetics
1992-	Canadian Angelman Syndrome Society
1993-	Founding Fellow in American College of Medical Genetics
1993-	Member, Prenatal Diagnosis Committee of New England Regional Genetics Group
1993-99	Cytogenetics Faculty Organizer and Member, Harvard Training Fellowship Program in Genetics
1994-	Member, Inverted Duplication 15 Education and Advocacy Group
1999-2004	National Committee for Clinical Laboratory Standards subcommittee advisor on Fluorescence In Situ Hybridization for Medical Genetics for FISH Validation (Report issued 2004)
2000, 2001	Laboratory Inspector, College of American Pathologists
2001-	Active Member, American Cancer Research Association

Major Research Interests:

1. Chromatin/chromosomal structure
2. Genetic imprinting of chromosome 15q11q13 in Angelman syndrome, Prader-Willi syndrome, and duplication syndromes
3. Phenotype/genotype relationships in human genetic disorders
4. Molecular mechanisms causing chromosomal rearrangements
5. Bioinformatics and development of new DNA technologies for detection of human genetic disorders

Principal Clinical and Hospital Service Responsibilities:

1991-1999	Scientific Director, Clinical Cytogenetics Service, Beth Israel Hospital, Boston
1999-	Director, Clinical Cytogenetics, Children's Mercy Hospital, Kansas City, MO

Teaching:

1992-99	Genetics Laboratory Tutorials: Demonstrator, first year Harvard medical students
1992-99	Cytogenetics lectures, Beth Israel Hospital: Lecturer, first through fourth year Pathology residents
1992-99	Cytogenetics Laboratory Rotations: Director of service, students, second year Pathology residents and Genetics Fellows in Harvard Training Program in Genetics, ~5 per year at one month each
1993-99	Cytogenetic and molecular genetics lectures for fellows in Harvard Training Program in Genetics
1996, 1997	Prenatal Diagnosis Lecture to students in HMS/MIT Reproductive Biology program
1999-	Management of Continuing Medical Education Lectures at Children's Mercy Hospitals
2001-	Clinical Cytogenetics, UMKC Medical Students (3rd Year)
2001-	Mentor/Molecular Biology and Cytogenetics Instructor to two Ph.D. Candidates from Kansas University's Department of Chemistry

Advising Responsibilities:

1994	Medical student in research laboratory (Nina Livingston).
1992-1993	Postdoctoral fellow in research laboratory. (Dr. Sou-De Chang who is currently head of Department of Anatomy. Chang Gung University, College of Medicine, Taipei Taiwan).
1993-99	Research technicians (Lisa White, Heather Baker).
1991-99	Clinical technologists (Alena Leff, Min Zhang, Amy Tillman, Shoshana York, JoAnn Rosol-Donoghue, Camille Marsh Scott).
1992-99	Clinical pathology residents rotating through the clinical laboratory
1992-99	Faculty, Harvard Genetics Training program
1995-97	Medical graduate student in research laboratory (Dr. Christine Mundlos).
1996-98	Clinical genetics fellow in research laboratory (Dr. Gabriella Repetto who is now director of clinical genetics services in University Catolica de Chile, Santiago, Chile).
1999-	16 clinical laboratory technical staff, 2 office assistants (minimal turnover in last 5 years).
1999-	2 research laboratory technical staff (Patrick Angell, Mauricio Miralles; previous: Angela Marion, Patricia Walters, Patricia Cazcarro, Brad Dalton), 0.5 office assistant (Ann Lowenstein; previous: Amy Wolfe).

Patents/Patent Applications:

Invention Disclosure 00-0001 (Children's Mercy Hospital); Selection and generation of single-copy genomic probes for hybridization; Disclosure Date: 3/24/00; Status: Patent application Ser. No. 09/573,080, filed 5/16/00; Allowed 8/13/03; Continuation application, filed 2/18/04. Patent #6,828,097 issued 12/08/04.

Invention Disclosure 01-0001 (Children's Mercy Hospital); Single copy probes and method of generating same (Continuation-in-part); Disclosure Date: 4/7/00; Status: US Patent App. Ser. No. 09/854,867, filed 5/14/01; PCT/US01/15674.

Invention Disclosure 01-0002 (Children's Mercy Hospital); Subtelomeric DNA probes and method of producing same; Disclosure Date: 9-15-02; Status: US Patent App. Ser. No. 60/415,345, filed 9/30/02. Improved Sub-telomeric DNA Probes and Method of Producing Same, filed 07/2/03. Subtelomeric DNA probes and method of producing same, PCT\US03\31170, WO 2004/029283 A2, US Patent App. Ser # 10\676,248, filed 9/30/03.

Invention Disclosure 01-0004 (Children's Mercy Hospital); Computational selection of probes for localizing chromosome breakpoints in genetic diseases and cancer; Disclosure Date: 4/15/04; Status: US Patent App. Ser #60/557,007, filed 3/26/04.

Bibliography:

Original Reports:

1. Knoll JHM. Frequency and replication pattern of fragile Xq28 in human heterozygotes from families with X-linked mental retardation. University of Saskatchewan, 1982.

2. Knoll JHM. Roberts Syndrome: Cytological and molecular investigations. University of Manitoba, 1987. (Ph.D. Thesis).

3. Chudley AE, Knoll JH, Gerrard JW, Shepel L, McGahey E, Anderson J. Fragile X-linked mental retardation I: Effect of age and intelligence on expression of the fragile X. Amer J Hum Genet 1983; 14:699-712.

4. Knoll JH, Chudley AE, Gerrard JW. Fragile X-linked mental retardation II: Frequency and replication

pattern of fragile Xq28 in heterozygotes. Amer J Hum Genet 1984; 36:640-645.

5. Chudley AE, Rozdilsky B, Houston CS, Becker LE, Knoll JH. Multicore disease in sibs with severe mental retardation, short stature, facial anomalies, hypoplasia of the pituitary fossa, and hypogonadatropic hypogonadism. Amer J Med Genet 1985; 20:145-158.

6. Hagerman RJ, Chudley AE, Knoll JH, Jackson AW, Kemper M, Ahmad R. Autism in fragile X females. Amer J Med Genet 1986; 23:375-380.

7. Heartlein MW, Knoll JHM, Latt SA. Chromosome instability associated with human alphoid DNA transfected into the Chinese hamster genome. Mol Cell Biol 1988; 8(9):3611-3618.

8. Knoll JHM, Nicholls RD, Magenis E, Graham JM Jr, Lalande M, Latt SA. Angelman and Prader-Willi syndromes share a common chromosome 15 deletion but differ in parental origin of the deletion. Amer J Med Genet 1989; 32(2):285-290.

9. Nicholls RD, Knoll JH, Glatt K, Hersh JH, Brewster TD, Graham JM Jr, Wurster-Hill D, Wharton R, Latt SA. Restriction fragment length polymorphisms within proximal 15q and their use in molecular cytogenetics and the Prader-Willi syndrome. Amer J Med Genet 1989; 33:66-77.

10. Knoll JHM, Nicholls RD, Lalande M. On the parental origin of chromosome 15q11q13 in Angelman Syndrome. Hum Genet 1989; 83:205-206.

11. Nicholls RD, Knoll JHM, Butler MG, Karam S, Lalande M. Genetic imprinting suggested by maternal heterodisomy in nondeletion Prader-Willi syndrome. Nature 1989; 342:281-285.

12. Knoll JHM, Nicholls RD, Magenis RE, Glatt K, Graham JM Jr, Kaplan L, Lalande M. Angelman Syndrome: Three molecular classes identified with chromosome 15q11q13 specific DNA markers. Amer J Hum Genet 1990; 47:149-155.

13. Bianchi DW, Flint AF, Pizzimenti MF, Knoll JHM, Latt SA. Isolation of fetal DNA from nucleated erythrocytes in maternal blood. Proc Nat Acad Sci 1990; 87(9):3279-3283.

14. Knoll JHM, Glatt K, Nicholls RD, Malcolm S, Lalande M. Chromosome 15 uniparental disomy is not frequent in Angelman Syndrome. Amer J Hum Genet 1991; 48:16-21.

15. Wagstaff J, Knoll JHM, Fleming J, Kirkness EF, Martin-Gallardo A, Greenberg F, Graham JM Jr, Menninger J, Ward D, Venter JC, Lalande M. Localization of the gene encoding the $GABA_A$ receptor B3 subunit to the Angelman/Prader-Willi region of human chromosome 15. Amer J Hum Genet 1991; 49:330-337.

16. Chaillet JR, Knoll JHM, Horsthemke B, Lalande M. The syntenic relationship between the critical deletion region for the Prader-Willi/Angelman syndromes and proximal mouse chromosome 7. Genomics 1991; 11:773-776.

17. Buiting K, Greger V, Horstmann I, Ludecke JJ, Senger G, Claussen U, Brownstein BH, Schlessinger D, Knoll JHM, Lalande M, Zabel B, Horsthemke B. Microdissection and molecular analysis of proximal 15q. In: Cassidy SB, ed. Prader-Willi syndrome and other chromosome 15q deletion disorders. Springer Verlag, Berlin. Series H: Cell Biology 1991; 61:13-17.

18. Beggs AH, Byers TJ, Knoll JHM, Boyce FM, Bruns G, Kunkel LM. Cloning and characterization of two human skeletal muscle alpha-actinin genes on chromosomes one and eleven. J Biol Chem 1992; 267:9281-9288.

19. Wagstaff J, Knoll JHM, Glatt KA, Shugart YY, Sommer A, Lalande M. Linkage of nondeletion Angelman syndrome to chromosome 1511-q13: Maternal but not paternal transmission leads to phenotypic expression. Nature Genetics 1992; 1:291-294.

20. Webb T, Clayton-Smith J, Cheng X-J, Knoll JHM, Lalande M, Pembrey ME, Malcolm S. Angelman syndrome with a chromosomal inversion 15 (p11q13) accompanied by a deletion in 15q11q13. J Med Genet 1992; 29:921-924.

21. Knoll JHM, Wagstaff J, Lalande M. Cytogenetic and molecular studies in Prader-Willi and Angelman syndromes. Amer J Med Genet 1993; 17:694-698.

22. Knoll JHM, Sinnett D, Wagstaff J, Glatt K, Wilcox AS, Whiting P, Wingrove P, Sikela JM, Lalande M. FISH ordering of reference markers and of the gene for the α5 subunit of the gamma-aminobutyric acid receptor (GABRA5) within the Angelman and Prader-Willi syndrome chromosome regions. Hum Molec Genet 1993; 2:183-189.

23. Warman ML, Tiller GE, Polumbo PA, Seldin MF, Rochelle JM, Knoll JHM, Cheng SD, Olsen BR. Physical and linkage mapping of the human and murine genes for the α1 chain of type IX collagen (COL9A1). Genomics 1993; 17:694-698.

24. Peters K, Knoll JHM. Diagnosis of tumors: The application of cytogenetics and fluorescence-in-situ-hybridization. Verh Dtsch Ges Zyt 1993; 18:66-68.

25. Oh SP, Warman P, Selden M, Cheng SD, Knoll JHM, Timmons S, Olsen BR. Cloning of cDNA and genomic DNA encoding human type XVIII collagen and localization of the α1(XVIII) collagen gene to mouse chromosome 10 and human chromosome 21. Genomics 1994; 19; 494-499.

26. Knoll JHM, Cheng SD, Lalande M. Allele specificity of DNA replication timing in the Angelman/Prader-Willi syndrome imprinted chromosomal region. Nature Genet 1994; 6:41-46.

27. Warman ML, McCarthy MT, Peralla M, Vubrio E, Knoll JHM, McDaniels CN, Mayne R, Beier DR, Olsen BR. The genes encoding alpha 2(ix) collagen (Col 9A2) map to human chromosome 1p32.3 to 1p33 and mouse chromosome 4. Genomics 1994; 23:158-162.

28. Shi GP, Webb AC, Foster KE, Knoll JHM, Lemere CA, Munger JS, Chapman HA. Human cathepsin S: chromosomal localization, gene structure and tissue distribution. J Biol Chem 1994: 23(1):158-162.

29. Cheng SD, Spinner NB, Zackai EH, Knoll JHM. Cytogenetic and molecular characterization inverted duplicated chromosomes 15 from eleven patients. Amer J Hum Genet 1994; 55:753-759.

30. Knoll JHM, Asamoah A, Pletcher BA, Wagstaff J. Interstitial duplication of proximal 22q: Phenotypic overlap with cat eye syndrome. Amer J Med Genet 1995; 55:221-224.

31. Mundlos S, Mulliken JB, Abramson DL, Warman ML, Knoll JHM, Olsen BR. Genetic mapping of cleidocranial dysplasia and evidence of a microdeletion in one family. Hum Mole Genet 1995; 4:71-75.

32. White L, Knoll JHM. Angelman syndrome: Routine molecular cytogenetic analysis of chromosome 15q11-q13. Amer J Med Genet 1995; 56:101-105.

33. Spinner NB, Zackai E, Cheng SD, Knoll JHM. Supernumerary inv dup(15) in a patient with Angelman syndrome and a deletion of 15q11-q13. Amer J Med Genet 1995; 57:61-65.

34. Saal K, Knoll JHM, Kadin M. Cytogenetic findings in regressing skin lesions of lymphomatoid papulosis. Canc Genet & Cytogenet 1995; 80:13-16.

35. Greger V, Knoll JHM, Woolf E, Glatt K, Tyndale RF, Olsen RS, Tobin AJ, SIkela JM, Nakatsu Y, Brilliant M, Whiting PJ, Lalande M. The τ-aminobutyric acid receptor τ3 subunit genet (GABRG3) is tightly linked to the α5 subunit gene (GABRA5) on human chromosome 15q11-q13 and is transcribed in the same orientation. Genomics 1995; 26:258-264.

36. Azim AC, Knoll JHM, Beggs AH, Chishti AH. Isoform cloning, actin binding, and chromosomal localization of human erythroid dematin, a member of the villin superfamily. J Biol Chem 1995; 270:17404-17413.

37. Simmons CF, Jr, Clancy TE, Quan R, Knoll JHM. The oxytocin receptor gene (OXTR) localizes to human chromosome 3p25 by fluorescence in situ hybridization and PCR analysis of somatic cell hybrids. Genomics 1995; 26:623-625.

38. Williams CA, Angelman H, Clayton-Smith J, Driscoll DJ, Hendrickson JE, Knoll JHM, Magenis RE, Schinzel A, Wagstaff J, Whidden EM, Zori RT. Angelman syndrome: Consensus for diagnostic criteria. Amer J Med Genet 1995; 56:237-238.

39. Schwartz F, Eisenman R, Knoll J, Gessler M, Bruns G. cDNA sequence, genomic organization and evolutionary conservation of a novel gene from the WAGR region. Genomics 1995; 29(2):526-532.

40. Azim AC, Knoll JHM, Marfatia SM, Peel DJ, Bryant PJ, Chishti AH. hDlg: Chromosome location of the closest human homologue of the Drosophila discs large tumor suppressor gene. Genomics 1996; in press.

41. Eksiolu YZ, Scheffer IE, Cargenas P, Knoll J, DiMario F, Ramsby G, Berg M, Kamuro K, Duyk GM, Huttenlocher PR, Walsh CA. Periventricular heterotopia: a novel X-linked dominant epilepsy gene in Xq28. Neuron 1996; 16:77-87.

42. Cassidy SB, Beaudet AL, Knoll JHM, Ledbetter DH, Nicholls RD, Schwartz, Butler MG, Watson M. Diagnostic testing for Prader-Willi and Angelman syndromes: Report of the ASHG/ACMG test and technology transfer committee. Amer J Hum Genet 1996; 58:1085-1088.

43. White L, Rogan PK, Nicholls RD, Wu B-L, Korf B, Knoll JHM. Allele-specific replication of 15q11q13 loci: A diagnostic test for detection of uniparental disomy. Amer J Hum Genet 1996; 59:423-430.

44. Vendetti CP, Knoll JHM, Sakthivel R, Harris JM, Chorney KA, Mowry PN, Schwartz S, Lacey PG, Phatak PD, Chorney MJ. 46,XX,inv(6)(p21.1p23) in a patient with idiopathic hemochromatosis (HFE): Relevance to the localization of the disease gene. J Med Genet 1997; 34:24-27.

45. Greger V, Knoll JHM, Wagstaff J, Woolf E, Lieske P, Glatt H, Benn PA, Rosengren SS, Lalande M. Angelman syndrome associated with a paracentric inversion 15 (q11.2q24.3). Amer J Hum Genet 1997;

60:574-580.
46. Engle EC, Castro AE, Macy ME, Knoll JHM, Beggs AH. A gene for isolated congenital ptosis maps to a 3-cM region within 1p32-p34.1. Amer J Hum Genet 1997;60-1150-1157.

47. Douhan J III, Lieberson R, Knoll JHM, Shou H, Glimcher LH. An isotype-specific activator of major histocompatibility complex (MHC) class II genes that is independent of class II transactivator. J Exp Med 1997; 185:1885-1895.

48. Gerecke DR, Olson PR, Koch M, Knoll JHM, Taylor R, Hudson DL, Champliaud MR, Olsen BR, Burgeson RE. Complete primary structure of two splice variants of human collagen XII, and assignment of alpha 1(XII) collagen (COL12A1), alpha 1 (IX) collagen (COL9A1) and alpha 1(XIX) collagen (COL19A1) to human chromosome 6q12-q14. Genomics 1997; 41(2):236-242.

49. Mundlos S, Otto F, Mundlos C, Mulliken JB, Aylsworth AS, Albright S, Lindout D, Cole WG, Henn W, Knoll JHM, Owen MJ, Zabel BU, Mertelsmann R, Olsen BY. Mutations involving the transcription factor CBFA1 cause cleidocranial dysplasia. Cell, 1997; 89(5):773-739, comment 677-680.

50. White LM, Treat K, Leff A, Styers D, Mitchell M, Knoll JHM. Exclusion of uniparental inheritance of chromosome 15 in a case with a familial dicentric (Y;15) translocation. Prenatal Diagnosis 1998; 18:111-116.

51. Repetto GR, Korf BR, Wagstaff J, Knoll JHM. Complex familial rearrangement of chromosome 9p24 detected by FISH. Amer J Med Genet, 1998; 76:306-309.

52. Blank V, Knoll JHM, Andrews NC. Molecular characterization and localization of the human MAFG gene. Genomics 1997; 44:147-149.

53. Kim AC, Peters LL, Knoll JHM, VanHuffel C, Ciciotte SL, Kleyn P, Chishti AH. Limatin (LIMAB1), an actin-binding LIM protein, maps to mouse chromosome 19 and human chromosome 10q25, a region frequently deleted in human cancers. Genomics 1997; 46:291-293.

54. Tiller GE, Warman ML, Gong Y, Knoll JHM, Mayne R, Brewton RG. Physical and linkage mapping of the gene for the alpha 3 chain of type IX collagen, COL9A3, to human chromosome 20q13.3. Cell Genetics and Cytogenetics 1998; in press.

55. Ott G, Katzenberger T, Siebert R, DeCoteau JF, Fletcher JA, Knoll JHM, Kalla J, Rosenwald A, Michaelason M, Ott MM, Weber-Matthiesen K, Kadin ME, Muller-Hermelink HK. Chromosomal abnormalities in nodal and extranodal CD30+ anaplastic large cell lymphomas: Infrequent detection of the t(2;5) in extranodal lymphomas. Genes, Chromosomes and Cancer 1998; 22:114-121.

56. Rogan PK, Seip JR, White LM, Wenger SL, Steele MW, Sperling MA, Menon R, Knoll JHM. Relaxation of imprinting in Prader-Willi syndrome. Human Genetics 1998: 103:694-701.

57. Sterpetti P, Hack A, Bashar M, Cheng SD, Knoll JHM, Toksoz D. Activation of the Lbc Rho Exchange Factor-Proto-oncogene by truncation of an extended C-terminus that regulates transformation and targeting. Mole Cell Bio 1999: 19(2); 1334-1345.

58. Repetto GM, White LM, Bader PJ, Johnson D, Knoll JHM. Interstitial duplication of proximal chromosome 15q in three patients with autistic features and mental retardation. Amer J Med Genet 1998; 79:82-89.

59. Hahn WC, Stewart SA, Brookls M, York SG, Ng-Eaton E, Kurachi A, Beijersbergen RL, Knoll JHM, Meyerson M, Weinberg RA. Inhibition of telomerase limits the growth of human cancer cells. Nature Medicine 1999; 5:1164-1170.

60. Kocher O, Comella N, Gilchrist A, Pal R, Tognazzi K, Brown LF, Knoll JHM. PDZK, a novel PDZ domain-containing protein upregulated in carcinomas and mapped to chromosome 1q21, interacts with cMOAT (MRPs), the multidrug resistance-associated protein. Lab Invest 1999; 79(9):1161-70 (issue cover).

61. Austin-Ward ED, Castillo S, Gragnic Y, Sanz P, Salazar S, Knoll JHM. Clinical findings in a patient with a supernumerary ring chromosome 20. Amer J Med Genet 91:171-174, 2000.

62. Ming JE, Blagowidow N, Knoll JH, Rollings L, Fortina P, McDonald-McGinn DM, Spinner NB, Zackai EH. A submicroscopic deletion in cousins with Prader-Willi syndrome causes a grandmatrilineal inheritance pattern: effects of imprinting. Amer J Med Genet 92:19-24, 2000.

63. Rogan, PK, Cazcarro, P, Knoll JHM. Sequence-based design of single-copy genomic DNA probes for fluorescence in situ hybridization. Genome Research 11: 1086-1094, 2001.

64. O'Sullivan, MJ, Swanson, PE, Knoll, J, Taboada, EM, Dehner, LP. Undifferentiated embryonal sarcoma with unusual features arising within mesenchymal hemartoma of the liver: Report of a case and review of the literature. Pediatr Dev Pathol. Sep-Oct 4(5): 482-489, 2001.

65. Yu N, Kruskall MS, Yunis JJ, Knoll JHM, Uhl, L, Alosco S, Ohashi M, Clavijo O, Husain Z, Yunis EJ, Yunis JJ, Yunis EJ. Disputed maternity leading to identification of tetragametic chimerism. New England J Med 346 (20): 1545-1552, 2002.

66. Knoll JHM, Rogan PK. Sequence-based, *in situ* detection of chromosomal abnormalities at high resolution, American Journal of Medical Genetics, 121A: 245-257, 2003.

67. Rogan PK, Knoll JHM. High resolution detection of chromosome abnormalities with single copy fluorescence in situ hybridization. IEEE Symposium on Biomedical Engineering, April, 2004.

68. Rogan PK, Knoll JHM: High Resolution Definition of Chromosome Abnormalities with Probes Designed from Genome Sequences, Discovery Medicine, 21(4): 99-101, 2004.

69. Tetsuya K, Shyu C, He L, Rogan PK, Knoll JHM. Content and classification based ranking algorithm for metaphase chromosome images. Proceedings of the IEEE International Conference, accepted for publication.

70. Knoll JHM, Marion AM, Fletjer W, Persons D, Cowan J, Rogan PK. Precise definition of chromosome 9 deletion and non-deletion breakage intervals in chronic myelogenous leukemia. Submitted 2004.

71. Knoll JHM, Angell P, Walters P, Marsh C, Rogan P. Generation of high-specificity, single-copy probes proximate to human telomeres. Submitted 2004.

Reviews and educationally relevant publications:

1. Knoll JHM, Wagstaff J, Lalande M. Prader-Willi Syndrome. In: Smith B, Adelman G, eds. Neuroscience Year: Supplement 3 to the Encyclopedia of Neuroscience. Birkhauser, Boston, MA. 1993; pp 132-133.

2. Lalande M, Wagstaff J, Knoll JHM. Molecular Analysis of the Angelman/Prader-Willi Syndromes. In: Adolph KW, ed. Genome Research in Molecular Medicine and Virology, New York, NY. Academic Press. 1993; pp 69-82.

3. Lalande M, Wagstaff J, Sinnett D, Greger V, Knoll JHM. Mapping of the Angelman and Prader-Willi syndromes. In Epstein CJ (ed): The phenotypic mapping of Down syndrome and other aneuploid conditions, 1993; 225-234, Wiley-Liss, New York.

4. Knoll JHM, Lichter P. In situ hybridization to metaphase chromosomes and interphase nuclei. In Dracopoli NC, Haines JL, Korf BR, Moir DT, Morton CC, Seidman CE, Seidman JG, Smith DR (eds): "Current protocols in Human Genetics Volume 1" 1994: Unit 4.3, Green-Wiley, New York. (Revised July, 2004).

5. Knoll JHM. Prader-Willi Syndrome. In: Smith B, Adelman G, Eds. Neuroscience Year: Supplement 4 to the *Encyclopedia of Neuroscience.* Birkhauser, Boston, MA. 1996.

6. Robinson, WP, Horsthemke B, Leonard S, Malcolm S, Morton CC, Nicholls RD, Ritchie R, Rogan P, Schultz R, Sxhwartz S, Sharp J, Trent R, Wevrick R, Williamson M, Knoll JHM. Report of the third international workshop on human chromosome 15 mapping 1996. Cytogenetics and Cell Genetics 76:1-13, 1997.

7. Knoll JHM. Genomic Imprinting. In: McGraw-Hill 1999 Yearbook of Science and Technology. December, 1998.

8. Knoll JHM. A Cytogenetic nightmare. Case 4 for Genetics, Embryology, and Reproduction for first year medical students, Harvard Medical School, 1997; 1998; 1999.

9. Knoll JHM, Rogan PK: Prader Willi Syndrome, Encyclopedia of Neuroscience, 3rd edition (George Adelman and Barry H. Smith, eds.), in press.

Abstracts (last 5 years):

1. The human SRA1 gene maps proximal to the Prader-Willi/Angelman syndrome domain in 15q11 and is non-imprinted. Amer J Hum Genet 65(4):A458, 1999.

2. JHM Knoll, H Baker, G Cox, ML Begleiter, LM Pasztor. Parental origin and replication timing studies in a 70, XXXX liveborn. Amer J Hum Genet 65(4):A167, 1999.

3. Kruskall MS, Uhl L, Yu N, Hussein S, Yunis E, Knoll J. Misinterpretation of HLA typing in a woman with congenital chimerism due to postzygotic fusion of two embryos. American Association of Blood Banking Meeting, November, 2000.

4. Knoll JHM, Cazcarro P, Rogan PK: Clinical application of sequence-based single copy probes for FISH, American Society of Human Genetics, October 2000.

5. Rogan PK, Cazcarro P, Knoll JHM. Single copy hybridization probes derived by genomic sequence analysis. American Society of Human Genetics, October 2000

6. Rogan PK, Cazcarro P, Knoll JHM: Single copy hybridization probes for detection of chromosome rearrangements derived by genomic sequence analysis. American Association for Cancer Research, March 2001.

7. Rogan PK, Cazcarro P, Knoll JHM: Single copy hybridization probes for detection of chromosome rearrangements derived by genomic sequence analysis. 10th International Congress of Human Genetics (platform), May 2001.

8. Rogan PK, Cazcarro P, Knoll JHM: Novel features of genome organization revealed by single copy FISH. American Society of Human Genetics, October 2001.

9. Cazcarro P, Rogan PK, Knoll JHM: Rapid sequence-based definition of chromosomal abnormalities. American Society of Human Genetics, October 2001.

10. Knoll JHM, Rogan PK: Detection of chromosomal rearrangements with sequence-defined, single copy hybridization probes. Second International NCI-EORTC Meeting on Cancer Diagnostics: From Discovery to Clinical Practice. June, 2002.

11. Knoll JHM, Angell P, Rogan PK: Detection of chromosomal rearrangements with single copy FISH probe arrays. American Society of Human Genetics, October 2002.

12. Knoll JHM, Rogan PK. Detection of Chromosomal Rearrangements with Single Copy FISH. 2nd Annual Kansas City Area Life Sciences Day, March 2003.

13. J.H.M. Knoll & P.K. Rogan. Detection of Chromosomal Rearrangements with Single Copy FISH. XIX International Congress of Genetics, July 2003.

14. Joan H.M. Knoll and Peter Rogan. Personalized Molecular Cytogenetic Diagnosis. 3rd Annual Kansas City Area Life Sciences Day, June 2004.

15. Knoll JHM, Marion AM, Fletjer W, Persons D, Cowan J, Rogan PK. Small upstream deletions of *ABL* 1 are rare in Chronic Myelogenous Leukemia. 2004 ASHG Annual Meeting, November, 2004.

16. Knoll JHM, Rogan P. Applications of personalized molecular cytogenetic diagnostics. 2004 ASHG Annual Meeting, November, 2004.

17. Yanala P., Lu T., El-Ghussein F., Zhao C., Medhi D., Wang Y-P., Knopp J., Knoll JHM., Rogan P. Automated detection of metaphase chromosomes in FISH and routine cytogenetics. 2004 ASHG Annual Meeting, November 2004.